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INTERDISCIPLINARY APPROACHES FOR TOTAL COMMUNICATION WITHIN DISABILITY SETTINGS (O11)

Co-funded by the Erasmus+ Programme of the European Union

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Interdisciplinary Approaches for Total Communication within Disability Settings (O11)

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Output 11 comprises a selection of documents contributed by researchers who participated in the Project International Conference and Multiplier Event, December 2017. These contributions have been selected by the scientific committee after a peer review process and they address important issues related to the EC+ Project.

There are five different blocks:

The first one, dedicated to intervention and communication, contains two chapters. The first addresses the topic of natural gestures for communication within the group of people suffering from severe intellectual disability and a second is dedicated to the communication with people suffering from Angelman syndrome.

The second block contains two chapters. The first one deals with the opportunities of ICT tools for communication and the second one with the specific application created by EC+ Project and its use for the intervention of people suffering profound intellectual disability.

The third block includes terminological resources for interpreters with a database for aphasia in the first chapter and a multilingual glossary for the 15 syndromes studied in the EC+ Project in the second chapter.

The fourth block contains two resources on multimodality approaches for deaf people using pictograms.

The last chapter is dedicated to legal and social aspects of disability and the present challenges.

1. Enhanced Natural Gestures: An Alternative to Natural Gestures and Sign Language for Individuals With Severe Disabilities

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Introduction

The primary purpose of this article is to provide an overview of Enhanced Natural Gestures (Calculator, 2002; Calculator, 2016; Calculator & Diaz-Caneja Sela, 2015) along with rationale for their use with individuals with severe disabilities. These are individuals who typically present with severe to profound intellectual delays that are accompanied by substantial challenges across a variety of other areas. These may include disorders of movement, sensory function (particularly vision and hearing), overall health, behavior, and communication.

Enhanced Natural Gestures (ENGs) are intended to be used in conjunction with other gestural methods, as well as vocal and graphic forms of communication. As such, they comprise one component of a multimodal communication system. They are considered one of many forms of augmentative and alternative communication (AAC). AAC becomes necessary when individuals with disabilities are unable to use speech as their primary means of communication.

Overview of Augmentative and Alternative Communication

AAC, including the use of ENGs, is usually intended to augment, or, supplement individuals' existing multimodal methods of communication. However, in some cases it is introduced to replace other undesirable methods. This is most often the case with respect to individuals' reliance on challenging behaviors such as hitting, spitting, and pulling hair in the absence of other more conventional and socially acceptable ways of communicating.

ENGs and other AAC methods may also serve as alternatives to existing methods of communication that are highly idiosyncratic, or ,specific to a particular individual and thus difficult to be interpreted and responded to correctly by other than a

select few (e.g. parents). This can include not only the use of natural gestures, described later, but sign language too.

ENGs can be an alternative to sign language in cases in which individuals' signs are substantially modified to accommodate intellectual, motor, and other limitations. As such signs may be recognizable to a limited audience. This may include communication partners who are well versed in sign language but unfamiliar with the modified and thus unique versions of signs produced by particular individuals.

The ensuing discussion of Enhanced Natural Gestures begins by examining their origin and how they are special relative to other forms of gestural communication, including natural gestures and sign language. This is followed by a brief review of research supporting the efficacy of ENGs as a communication method for individuals with severe disabilities. Next, teaching methods comprising the ENG protocols are described. These include discussions of two methods associated with incidental teaching: mand-model with time delay and molding-shaping. In both cases, all instruction is carried out in conjunction with normally occurring events through the use of incidental teaching. Next, the seven steps of the ENG program are reviewed briefly. Finally, implications of this evidence-based method are discussed as they pertain to maximizing the effectiveness with which individuals with severe disabilities communicate with others.

Origin of Enhanced Natural Gestures

This method of communication was initially developed for individuals with Angelman Syndrome (AS), a neurodevelopmental genetic disorder caused by deficient expression of the maternally derived UBE3A gene on the 15th chromosome. Angelman Syndrome occurs at an incidence of approximately 1 in 12,000 live births. It is associated with a broad range of disabilities that include severe to profound intellectual delays, seizures, motoric challenges, absence of speech, severe delays in both expressive and receptive language, etc. Anecdotal evidence (e.g. written and electronic correspondence as well as verbal reports from parents and practitioners) suggests ENGs are now being used in the USA and a broad with individuals whose severe disabilities can be attributed to a variety of other etiologies as well.

Multimodal Communication

The communicative challenges of individuals with severe disabilities are usually observed across all modes of communication. Their *vocal* behavior is characterized by

severe limitations or absence of speech. *Gesturally*, we typically see limited abilities to learn sign language due to intellectual and motor disorders that influence their acquisition and accuracy of signs. Finally, limitations in the area of *graphic* communication narrow options with respect to the complexity and variety of both electronic (e.g. speech generating devices [SGDs]) and nonelectronic (e.g. communication boards and communication books) methods of communication that might be considered.

Individuals with severe disabilities often favor methods of communication such as non-speech vocalizations, physical manipulations of people and objects, and natural gestures (Calculator, 2014). The latter are not taught but instead reflect self-devised behaviors such as pointing and reaching toward desired objects and events, and pushing way, avoiding, or withdrawing from unwanted objects and events.

Unfortunately the communicative behaviors of individuals with severe disabilities are usually severely restricted in number as well as likelihood of being understood by their conversational partners. Enhanced natural gestures were created with this is mind.

Characteristics of Enhanced Natural Gestures (ENGs)

Unlike natural gestures, which can be highly idiosyncratic and thus difficult for others to interpret, ENGs are by definition readily understood. Prior to being selected, the practitioner, which can include parents, siblings, teachers, and peers, presents a potential ENG to three naïve listeners, such as classmates or co-workers, along with the context in which it might arise. No fewer than two of these prospective partners must be able to identify the meaning of the ENG in order for it to be selected and then taught.

Thus, ENGs rarely need to be taught to familiar or unfamiliar conversational partners since they are highly transparent (i.e. guessable). This contrasts with sign language. Although some signs are transparent and easily interpreted, a large percentage are unlikely to be understood by anyone other than those with sign language training. In addition, due to individuals' motor limitations, their signs are often inaccurate and or highly modified. As such even unfamiliar conversational partners with backgrounds in sign language may be unable to interpret their meaning unless translated by others.

Unlike sign language, ENGs build upon natural gestures individuals are already displaying; thus they are easily taught and acquired. For example, an individual might be observed using both hands to lift and then bring a cup to his mouth to drink. This

same gesture (i.e. cupping both hands and bringing them to his mouth) could be taught as a means by which that individual might request a drink in its absence. Conversely, another individual might drink by grasping a cup with one hand before bringing it to her mouth. Her ENG would thus reflect this different behavior. As another example, an individual might be observed naturally pulling on the chains of a playground swing to propel herself. We might later observe this individual using an ENG (i.e. producing the same grasping and pulling motion) to request to be taken to a swing on the playground as a preferred activity. Finally, based upon the hand motions of an individual when swimming, we might extract this same gesture and teach it as an ENG for asking her parents to go outside and take a swim in the family pool.

Efficacy of Enhanced Natural Gestures

As indicated earlier, research has supported the efficacy of ENGs.Parents have reported them to be an effective, acceptable and feasible method of communication for their children. This has been confirmed in two home-based studies in which parents were taught to use ENGs and then asked to monitor their children's uses of ENGs over time (Calculator, 2002; 2018) and a third study in which educators were taught and then used ENGs with their students in school (Calculator & Diaz-Caneja Sela, 2015). Readers are encouraged to review these three studies in order to gain a comprehensive understanding of this method.

Teaching Methods

The studies cited above offer step-by-step descriptions of the ENG instructional protocols, which include tutorials on the two teaching strategies (i.e. mand-model with time delay and molding-shaping) that can be used alone or in combination. Both of these methods rely on incidental teaching. In other words, instructors take advantage of existing opportunities for communication that arise in the natural environment and/or structure natural environments to create reasons for individuals to use their ENGs.

Teaching strategies. Briefly, the first teaching method, mand-model with time delay, begins with the instructor emitting an expectant gaze to signal to the individual a need to use an ENG. It has already been established that a communicative behavior is expected to arise in this predictable, frequently occurring situation. The ENGis embedded in this event. If the individual fails to produce the ENG, the instructor mands (i.e. says "show me what you want," or "show me your gesture"). If the individual still fails to produce the ENG, the instructor models the desired ENG. If even then the

individual does not produce the desired ENG, the instructor physically prompts the individual to do so. This often requires full hand-over-hand assistance.

The second teaching strategy involves molding-shaping. First, the individual's hands are molded around a targeted object and she is encouraged to handle it in a customary way (e.g. grasp and then bounce a ball; grasp and then bring food to her mouth; grasp and then use sticks to beat a drum; etc.). Next, we move to the shaping phase. The object (e.g. ball) is removed from the individual's hands. She is encouraged to maintain the same hand position, as if still retaining possession of the object (e.g. the ball) and then execute the expected movement. If she fails to do so, the instructor repeats the molding and shaping phases already described. If the individual still fails to produce the targeted ENG, the instructor provides physical hand-over-hand assistance to prompt the individual to produce the ENG in the absence of the corresponding object.

Once again, readers are strongly encouraged to review the author's investigations that were cited earlier for additional information pertaining to the means by which ENGs are taught. This includes what has now evolved into a seven-stepprogram, which is summarized below.

Seven Steps for Teaching Enhanced Natural Gestures

In the first step, the instructorobserves the individual's existing methods of communication and evaluates their corresponding success. The resulting data are used as a means of determining candidacy for ENG training. For example, if the individual is already communicating successfully with a broad range of conversational partners, she may not be a candidate for ENG instruction.

In step two, we identify the situations or environments in which ENGs will be taught. These vary greatly. Common situations include mealtime, recess (time on the playground), music, art, bath-time,free play, work, and leisure activities. It is often recommended to begin with no more than three situations. These can be increased over time.

Step three requires the instructor to observe and rate the success with which the individual is already communicating in each of the situations identified earlier. Where reasons and opportunities for communication exist but communicative attempts are either absent or unsuccessful, there may be a basis for teaching ENGs as augmentative and/or alternative methods of communication.

Next, in step four, the actual ENGs are determined. As noted earlier, ENGs often involve replacements of existing natural gestures that may or may not be

idiosyncratic in nature. These may include undesirable challenging behaviors such as hitting. The transparency of potential ENGs is confirmed by previewing them with naïve conversational partners and confirming their guessability in context. No fewer than two out of three partners must be able to interpret each ENG successfully in order for it to be subsequently taught.

Step five involves the instructor and his/her team reaching consensus on a set of goals and expected outcomes of ENG instruction through the use of goal attainment scaling, or, GAS (Calculator, 2016; Kiresuk& Sherman, 1968; McDougall & King, 2007; Schlosser, 2004). One example of a goal identified by a team was that a student, John, would "use his ENGs in conjunction with other AAC methods to initiate no fewer than 10 interaction with his peers each day."Other goals have targeted the use of ENGs to request desired objects, request continuation of pleasurable activities, reject unwanted objects, request preferred objects, and request objects necessary to participate in a corresponding activity. For each goal, the instructor (or team) identifies a set of five possible outcomes: +2 (best expected). +1 (more than expected), 0 (expected), -1 (less than expected), or -2 (worst expected).

In step six, ENG instruction is implemented and data on the individual's performance in each situation are collected. As noted earlier, ENG instruction is fully integrated into each situation. For example, during shared storybook reading, a classmate (previously mentored by the classroom teacher on implementation of the ENG protocol) of Megan's who typically reads to her approached Megan and gazed expectantly. Based on previous experiences, Megan expected her classmate to have arrived with a book and commenced reading to her, but this was not the case on this occasion. Megan produced the ENG to request a book from her classmate. Megan's ENG was producing the hand shape associated with holding a book in her two hands. Megan's successful use of the ENG prompted her classmate to retrieve a book. In this case, the goal was for Megan to request an object needed to participate in an activity.

Another example of an event arising in step six involvedNoah. One of his goals was to have him request a preferred activity. Upon entering the music room, Noah expected to be escorted to the piano. However in this phase of instruction his teacher paused with Noah at the threshold of the room. Molding-shaping had been used to teach Noah to assume the hand shape and execute the movement associated with playing the piano. When confronted with the situation in which this preferred activity

was not immediately available, Noah produced the ENG for piano to communicate his want/need.

The final, seventh, stage of the program involves the instructor and/or team evaluating the efficacy of the ENG program and achievements of the individual. This is accomplished first by their completing the Enhanced Natural Gestures Acceptability Rating Form (see previous sources for a description of this tool). The instructor uses a series of Likert rating scales, along with responses to open-ended questions, to accomplish this. In addition, the goal attainment scaling procedure is completed in order to evaluate individuals' actual outcomes relative to expected outcomes. These can then be evaluated statistically.

Summary

The ENG approach has undergone several revisions since its introduction in 2002. These have been based on empirical data as well as anecdotal reports from practitioners, including parents and teachers. This feedback has alluded to features such as clarity, acceptability, reasonableness and time required to learn and then implement the ENG instructional protocols.

As noted above, three investigations have validated the efficacy of Enhanced Natural Gestures as a method by which individuals can be taught to communicate more effectively.In two of these investigations, one involving parents, the otherteachers (and otherschool personnel), efficacy was evaluated only through completion ofthe Enhanced Natural Gestures Acceptability Rating Form. The third investigation, involving parents, again relied on the ENGARF but this time in conjunction with Goal Attainment Scaling. The current iteration of the ENG instructional protocol embeds Goal Attainment Scaling into the program.

ENGs are learned and then used most effectively through an integrated approach in which instruction is embedded into everyday activities. Based on the research cited earlier, ENGs may be considered a viable alternative to natural gestures and other idiosyncratic methods of communication, which may include sign language. This may be especially true when ENGs comprise one component of a multimodal AAC system. ENGs should never be introduced as a means of replacing methods of communication that are already effective with a broad range of familiar as well as unfamiliar conversational partners. Further research is necessary to evaluate the degree to which ENGs can be effective with the broad population of individuals with severe disabilities.

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2. Hacia un perfil de Comunicación y Lenguaje en personas con Síndrome de Angelman

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Los fenotipos conductuales son comportamientos asociados a un genotipo, observados en personas con un síndrome genético específico. Las evidencias señalan la determinación orgánica de algunos comportamientos en niños con trastornos mentales incapacitantes de base biológica (Waite et. al, 2014; O´Brien, 2000).

El Síndrome de Angelman (SA) es un modelo perfecto de impronta genómica porque la información genética se expresa de manera diferente en función de la matriz de origen. Cuando los mecanismos que regulan la impronta del cromosoma 15q11 – q13 proceden de la madre origina el SA, afectando a 1:20000/30000 personas. Para Brun (2005) la frecuencia del diagnóstico en España se ha incrementado, el diagnóstico es más temprano debido tanto al perfeccionamiento de las pruebas diagnósticas como al mayor conocimiento de la patología.

El fenotipo se caracteriza por una apariencia feliz, fascinación por el agua, conducta hiperactiva, personalidad excitable, déficit de atención, ataques de risa, hipersensibilidad al calor y conductas masticatorias entre otros (Brun, 2000). Se mencionan cinco mecanismos genéticos pero todos comparten el retraso severo del desarrollo, las dificultades del habla, los trastornos en el movimiento y el equilibrio, el déficit cognitivo y la risa inmotivada:

1. *Deleción intersticial de novo:* en el cromosoma 15q11 – q13, con frecuencia del 65 – 70%. Fenotipo clásico y severo, hipopigmentación y convulsiones tempranas y generalmente graves.
2. *Disomía uniparental paterna*: heredan dos copias del cromosoma paterno y por impronta tiene los genes de la región SA silenciados. Frecuencia alrededor del 5%, menos severa que la deleción.
3. *Mutación de impronta*: los alelos de la madre están silenciados y no se expresan los genes de la región SA, frecuencia menor al 5%.

4. *Mutación intragénica del gen UBE3A*: síntesis inactiva de proteínas cerebrales que intervienen en la degradación de otras proteínas. Ocurre en 10% de casos familiares de metilación normal del ADN.
5. *Etiología desconocida*: Incluye al 10% de personas negativas a la mutación *UBE3A.*

Tempranamente se observa en la persona con SA dificultades de alimentación (succión débil, reflujo gastroesofágico, poca ganancia de peso a pesar de que lacta por largos períodos) y ausencia de llanto. Se evidencia un retraso en el desarrollo motor y una incapacidad para sentarse, el arrastre es común y en la mayoría de casos caminan independientemente alrededor de los 3 o 4 años. Una minoría no logra la locomoción y tienden a tener el mayor grado de espasticidad en extremidades u otras dificultades como la escoliosis. La marcha en el SA es característicamente atáxica de amplia base y piernas rígidas, los brazos apoyados en el codo (Clayton – Smith, 1992). Entre los rasgos físicos característicos destacan: lengua prominente, hipopigmentación, boca grande, dientes separados, microcefalia, manos y pies pequeños, braquicefalia con occipucio plano. El inicio de las convulsiones ocurre antes de los 3 años en el 80% de las personas. Entre los trastornos del movimiento se mencionan temblores, sacudidas y ataxia (Thompson & Bolton, 2003).

Las severas dificultades de comunicación en personas con SA manifiesta un fenotipo con ausente lenguaje funcional. En la comunicación no verbal predominan vocalizaciones, señales y gestos; y no se identifican en su vocabulario más de dos o tres palabras simples. Hyppa et. al (2013) señala un repertorio por debajo de los 10 signos y una minoría emplea comunicación gráfica; aunque señala idóneas las modalidades de comunicación gráfica y gestual en personas con SA. Entre las funciones de la comunicación menciona emplear conductas de demanda y protesta; y medios no simbólicos (alcanzar, acercarse, protestar, aleteo, llanto, risa).

Robb et al. (1989) revisó casos con SA entre 18 meses - 13 años, y señala DI severa en la totalidad y un déficit profundo de lenguaje, algunos decían una palabra hasta un máximo de tres. La evaluación de Jolleff & Ryan (1993) a personas con SA de hasta 15 años de edad, señala niveles de comprensión de 9 a 22 meses, no comprenden oraciones de más de 2 palabras simples y un habla expresiva entre 6 a 12 meses, la mayoría sin desarrollo de palabra alguna. Como habilidades de comunicación pre verbal menciona la utilización de manos y brazos para expresar necesidades; entre los

gestos y señales de comunicación no verbal prefieren guiar de la mano al adulto hacia lo que desean; la manipulación, canales físicos de comunicación y la mirada referencial. Las dificultades de planificación y ejecución motora incluyen al habla y el uso de gestos y otros signos no verbales; no consiguen imitar el aplauso o señales de saludo/despedida con la mano.

Entre las estrategias de intervención en personas con SA se mencionan: la enseñanza del uso de gestos naturales, estrategias de comunicación receptiva y seguimiento de instrucciones, comunicación expresiva a través de modos gráficos, entre otros. Hyppa et. al. (2013) reportaron una intervención que enfatiza la comunicación vocal, gestual y gráfica; enfatizaron la importancia de estas estrategias en las primeras etapas de intervención de comunicación aumentativa y evidenciando un mejor aprendizaje para solicitar artículos preferidos en modalidades de comunicación gráfica. Calculator (2013) señala una comunicación multimodal en personas con SA, que será más efectiva si se fomenta una comunicación completa, utilizando Sistemas de Comunicación Alternativa y Aumentativa (SAAC). Las personas con SA dependen de métodos de comunicación asistida o no, los no asistidos incluyen vocalizaciones no verbales, gestos naturales, conductas de demanda y signos; y su utilización temprana los introduciría a los dispositivos electrónicos. Sin duda, las personas con SA podrían comunicarse plenamente con la participación de la familia, y el uso de SAAC repercutiría en una mejora de la calidad de vida a corto y largo plazo (Calculator, 2010).

Objetivo:

Esta propuesta tiene como finalidad aproximarse a un perfil de lenguaje y comunicación en personas con SA según tipo de alteración genética.

Metodología:

Para ello se contactó con la Asociación de personas con Síndrome de Angelman (ASA), y se contactó a 31 padres o cuidadores. Según etiología genética, en 16 personas la causa fue deleción, en 5 disomía uniparental paterna, 4 por alteración del imprinting, 4 por mutación UBE3A y 2 con estudio molecular normal.

Se utilizó el Cuestionario 1 de la adaptación española de los Inventarios de Desarrollo Comunicativo de MacArthur (López Ornat S, Gallego C, Gallo P, Karousou A, Mariscal S., Martinez, M, 2005), que son inventarios dirigidos a padres de niños de 8 a 30 meses de edad. Las preguntas del Cuadernillo 1 contemplan el desarrollo

comunicativo como comprensión de frases y palabras, producción de palabras, gestos y acciones y uso del lenguaje; a diferencias del cuadernillo 2 que contempla un desarrollo léxico, gramatical y morfosintáctico de palabras y oraciones. Asimismo, los padres ofrecieron datos demográficos de la familia, uso de SAAC, estimulación logopédica.

Resultados:

Los resultados encontrados nos permiten una descripción del perfil de lenguaje y comunicación de personas con SA, de la siguiente manera:

- Conductas prelingüísticas correspondientes a 12 meses con predominio de vocalizaciones, gestos protoimperativos y protodeclarativos.
- Comprensión de frases de saludo y expresiones cotidianas en un nivel cercano a 15 meses aproximadamente.
- Vocabulario comprensivo cercano a 13 meses de edad, con mayor desarrollo en las categorías de rutinas y fórmulas sociales, personas entre otros.
- Vocabulario expresivo correspondiente a 15 meses, con mayor desarrollo en las categorías de personas, alimentos y bebidas.

Encontramos conductas prelingüísticas como utilizar gestos para pedir cosas, llamar la atención, repetir sílabas o vocalizaciones, intentos de imitar palabras y vocalizaciones y uso de vocalizaciones parecidas a palabras verdaderas. Entre las conductas de comprensión temprana encontramos, respuestas al llamado por su nombre, al *"no"* dejan de hacer y comprenden con la mirada si otros llaman a papá o mamá.

En comprensión global de frases; comprenden expresiones cotidianas y rutinarias. Los gestos más frecuentes encontrados son: extender los brazos para mostrar, alcanzar algún juguete a enseñar, levantar los brazos para levantar, hacer gesto de "*no"* con la cabeza. Como acciones: beber de taza, comer con cuchara, ponerse el teléfono en la oreja.

En relación al tipo de alteración genética causante del SA encontramos que las personas con SA por deleción obtienen peor resultado, seguido de las personas con estudio molecular normal. Finalmente, no observamos diferencias según la escolaridad de los padres, pero familias con padres sin escolaridad obtienen medias inferiores en algunos subtests explorados. En relación a las formas de comunicación y las terapias

especializadas, encontramos utilización de gestos naturales, tarjetas de imágenes, vocalizaciones, lenguaje de signos y unos pocos utilizan dispositivos electrónicos.

Discusión:

Los resultados resaltan características prelingüísticas correspondientes a vocalizaciones, gestos protoimperativos y acciones; Thompson & Bolton (2003) utilizaron las Escalas de Conducta Adaptativa de Vineland y obtuvieron niveles de 16 meses en el dominio de comunicación; utilizando el Inventario para la planificación de servicios y Programación Individual – ICAP Brun et. al (2010) señalaron niveles medios de 10 meses en destrezas sociales y comunicativas. Se han descrito habilidades similares de comunicación pre verbal en personas con SA, con predominio de gestos motores presimbólicos, vocalizaciones, ecolalia, conductas de demanda, canales físicos de comunicación y mirada referencial (Calculator, 2013; Didden et al., 2004; Hyppa et al, 2013; Thompson &Bolton, 2003). Se señala también un menor desarrollo a nivel de planificación y ejecución motora, señales, conductas imitativas, dificultades en rutinas y conductas de autonomía (Brun, 2005; Hyppa et. al, 2013; Jolleff & Ryan, 1993).

En relación a los mecanismos genéticos causantes del SA, la gran mayoría de casos se asocia a microdeleción 15q11- q13 con mayores defectos que la impronta o la disomía uniparental; y un 15% aproximadamente sin causa citogenética conocida. Las alteraciones moleculares por deleción, disomía uniparental, imprinting, *UBE3A* y estudio molecular normal ocurre en frecuencias del 70–75%, 2-5%, 1-5%, 10% y 10% respectivamente (Lalande & Calciano, 2007; Brun & Artigas, 2005), lo que se corresponde aproximadamente con nuestros resultados. La literatura reporta la correlación genotipo – fenotipo en aspectos lingüísticos (Lalande et al., 2007, Thompson et al., 2003; Clayton – Smith, 1992, Brun & Artigas, 2005; Gentile et al., 2010) al señalarse diferencias entre clases moleculares, en ese mismo sentido la clase molecular es un factor que contribuye a la correlación genotipo – fenotipo. Existen estudios que señalan la gravedad clínica relacionada a otras variables como la gravedad de la epilepsia, los trastorno del sueño y nivel de discapacidad intelectual (Brun & Artigas, 2010; Gentile et al., 2010). De allí la importancia de un estudio clínico molecular completo, porque permitiría ofrecer consejo genético y valor pronóstico de la enfermedad.

Conclusiones

El SA ocasiona DI severa y características de comunicación no simbólica con probables habilidades de comprensión que mejoran a las de expresión. Encontramos asociación con el genotipo, dónde las causas por deleción y no conocido obtienen peores niveles de desarrollo pre lingüístico y lingüístico que personas con SA por no deleción. También existirían diferencias culturales de contexto familiar y educativo que podrían influir en el nivel de desarrollo de la comunicación y del lenguaje de las personas con SA. En este sentido nos unimos al llamado a las familias, educadores y profesionales en la comunicación a identificar métodos y estrategias efectivos y con beneficios óptimos para comunicarse con personas con SA.

Limitaciones de la investigación

La metodología no contempla factores adicionales no contemplados en nuestro estudio y que podrían influir en los resultados (nivel de DI, frecuencia y aparición de crisis epilépticas, comorbilidad, etc.).

Direcciones futuras de investigación

Las investigaciones futuras podrían explorar el entorno cultural y familiar de las personas con SA, relacionando la calidad de vida, soporte familiar, estrategias, creencias y expectativas con las habilidades comunicacionales de las personas con SA. Sería de utilidad explorar las diferencias lingüísticas en las diferentes etapas del desarrollo y la perspectiva familiar en este sentido. Finalmente, sería interesante explorar el ambiente escolar, porque el aula varía en relación al hogar, y el contexto podría brindarnos información útil; así como el uso de sistemas de comunicación aumentativa y experiencias de intervención.

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3. Opportunities of Information and Communication Technologies in Total Communication

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Introduction

According to the European Committee on Equality and Non-discrimination, there are more than 80 million people with disabilities in Europe and this number is expected to increase in the future[1]. Most of the vast literature available on disabilities is focused on people with some capacities at a certain point. Some authors agree that there is a need to provide resources to people with severe disabilities and special communications needs (Light & McNaughton, 2014, p.107; Mirenda, 2014, pp. 19-27). Resources like Makaton (Vinales, 2013), PECS (Bondy& Frost, 2002) and total communication (Schlesinger, 1986) are valid alternatives for communicating with people with severe disabilities, but, frequently, they are not easily available when there is a communication need.

Nowadays, it is very common to have a smartphone in our pockets or a tablet at home or in our work place. Thus, a mobile application containing all the resources to help in an effective communication with severely intellectually or developmentally impaired people would be a valuable tool for families, caregivers, leisure assistants, teachers, healthcare workers, and community interpreters. They can simply install such a tool and search for a word they want to use in their communication.

Mobile devices are an example of Information and Communication Technology (ICT) that are effective and useful for multimodal or Total Communication. However, other ICT tools, like desktop computers and laptops can also be useful. Multimedia resources covering the multiple modes of communication can be available in the form of desktop or Web application, as well as mobile device application. We focus, however, on mobile devices because they are portable devices that we can bring with us all the time. Thus, it is more convenient to use in most of the cases compared to a desktop or laptop computer that is a heavy device that does not fit in the pockets.

[1]See http://assembly.coe.int/nw/xml/XRef/Xref-XML2HTML-en.asp?fileid=21339&lang=en (accessed in November 2017).

In this document, we discuss a set of advantages and limitations of the mobile devices for total communication (Section 2). After a brief review of some applications for these devices in Section 3, we focus on one particular application in Section 4: EC+, a tool developed in the context of an Erasmus+ project where the authors collaborate as developers. In Section 5, some potential future lines of research in the context of ICT for total communication are outlined and Section 6 concludes the document.

Advantages and limitations of mobile devices for total communication

A mobile device is a small computer. In spite of their size, there are mobile phones today that are more powerful than desktop computers. They have a powerful microprocessor and enough memory to run complex programs and applications.

We can find many kinds of mobile devices in the market. The most common examples are the mobile phone and the tablet, but there are some others. For example, smart watches are devices with the size of a watch that send notifications to the user and gather some information from their sensors, like the pulse of the user, the amount of oxygen in the blood, etc. Some other examples of mobile devices are Personal Digital Assistants (PDAs), pocket calculators, digital photographic cameras, portable gaming consoles, CPU sticks, digital camcorders and so on.

In the context of Total Communication, the most appropriate mobile devices are the smart phones and the tablets because they have a screen large enough to visualize pictures and videos, and a powerful microprocessor to run applications.

The evolution of mobile phones in the last decades has been very fast. The first phones in the 1970s were big devices only able to do phone calls. With the advances of circuit integration and miniaturization, the size of mobile phones was reduced until 2003 (approximately). However, the trend changed completely in the mid 2000s, due to the change in the habits of the users and the popularization of the touch screens in mobile phones (see Figure 13-1). In effect, the increase in the power of the microprocessors for mobile phones allowed the use of the devices as multimedia centers, the access to Internet, and the use of new apps to assist the user in their daily life, thus replacing the old PDAs. On the other hand, the touch screen replaced the keyboard and the only interface with the user in a mobile phone was the screen. The higher the screen, the easier to use. These facts explain the increase in the size of the mobile phones.

Regarding tablets, although they existed since the beginning of the XXI century, an important event in their popularization was the launch of the iPad by Apple in 2010. The tablet replaced the paper in many activities, for example, in the health domain, where the doctors could use a tablet to visit their patients and keep some notes about him/her, or the sales representative of a company, who had the opportunity to show his/her clients multimedia resources showing the product.

When talking about mobile devices, an important aspect to take into account is the Operating Systems that controls the device. An Operating System is a piece of software that offers an abstraction layer to the developers of the mobile devices, hiding the details of the hardware of the device. There are many Operating Systems for mobile devices. However, there are two that cover the 99.7% of the mobile devices in the market. The most popular is Android, installed in 85.0% of the mobile devices; and it is followed by iOS, installed in 14.7% of the mobile devices[2]. The other 0.3% of the mobile devices have another operative system like Windows Phone, Bada, Blackberry OS, Tizen, Symbian OS, etc.

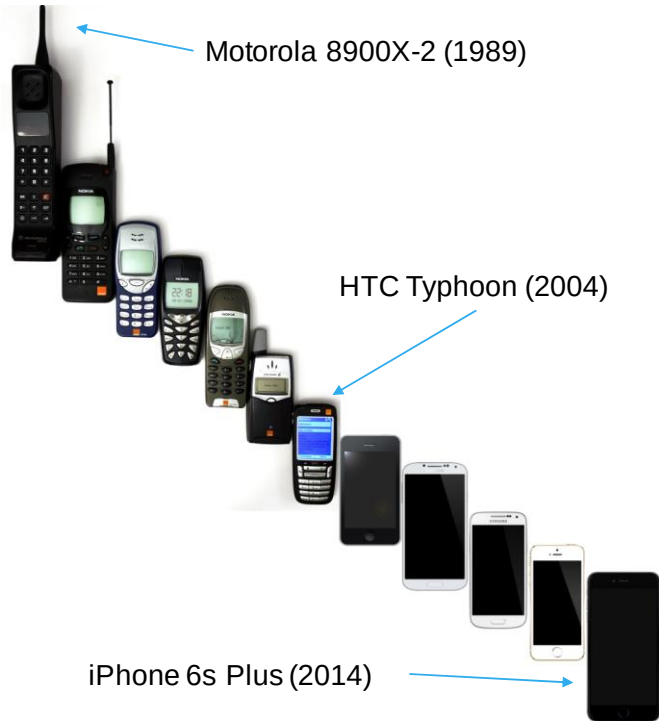

Figure 13 1: Evolution of mobile phones.

From the point of view of the developer, there are big differences between Android and iOS. While the former is completely open (the source code is available) and many resources and hints are available out there, the operative system of Apple is

[2]Source: IDC, http://www.idc.com/promo/smartphone-market-share/os

closed, there are more requirements to run a mobile application in a device and it requires a more expensive developer subscription than Android.

Independently of the Operating System installed in the device, there is a set of advantages and limitations that mobile devices have when they are used as tools in Total Communication. In the next lines, we will discuss these advantages and limitations.

Advantages

We identify six advantages of using mobile devices for total communication. They are the following:

1. Portability: the mobile devices can be taken with the user because they have a reduced weight and size. This contrasts with the computers of a few year ago, which were quite heavy and they could not be easily transported. Nowadays, we can have a device with a great computation power in the pocket.
2. Versatility: since the mobile phones have a general-purpose processor, they have a great versatility. The user can run applications of any kind, not only communication applications. For example, they can be smart applications that can act depending on the sensors using Artificial Intelligence; they can connect to the Internet to communicate with other devices and applications. This offers a great opportunity to develop smart applications for people with special communication needs.
3. Openness: this means that anybody can develop applications for mobile devices. In the case of iOS it is more difficult, but in Android, using App Inventor it is easy to develop a small application and install it in the mobile device.
4. Always with us: we usually have the mobile phone with us because it is a communication tool that we use to talk to the family and friends, using phone calls or text messages. Thus, if we need a special communication with a person with special needs, we do not need to use a special tool; we can simply take the phone from our pocket and use the corresponding application.
5. Communication tool: as a communication tool, the mobile phone can be used to communicate with a person in front of us or with a person which is thousands of kilometers apart. Thus, total communication can be used with persons far away.
6. Sensors: nowadays the mobile devices bring many sensors. Many of them are related with health (steps counter, oxygen in the blood, etc.). All these sensors of

the mobile phones can be used to create new modes of communication in total communication. Perhaps some persons cannot communicate the health state, but the mobile phone can do it on his/her behalf.

Limitations

However, the mobile devices also have limitations for total communication, and the ones identified are:

1. Battery: since the mobile device cannot be connected to the electric grid, it has a battery that provide the power, and it reduces the charge as time passes. As the computation power and the size of the screen of the devices is higher, the battery charge is exhausted earlier. The mobile devices of a few years ago could be working with a battery charge during a week, but mobile devices nowadays require a daily charge.
2. Memory: the memory is limited in mobile devices. The typical value for most of the devices today is 32GB, while in the case of computers the storage space is around 1TB (1 thousand GBs). The developers have to carefully think how to manage this small amount of memory to be able to run all the applications in the memory of the device.
3. Computation power: in the mobile phones, the processors have a lower performance, compared to the desktop processors. Developers have to take into account this fact to avoid that the battery charge is quickly exhausted. They also have to check if the mobile device is ready for a computationally costly task, because it could affect the temperature of the processor and the live of the mobile devices itself.
4. Screen: the size of the screen is also limited. There are things that are not appropriate in a mobile screen. For example, an application with many controls (buttons, for example) is not comfortable to user.
5. Bandwidth and coverage: the bandwidth of mobile devices is limited compared to other devices, like a desktop computer. The application in the mobile phone has to take into account this limited bandwidth to avoid a bad experience for the user. On the other hand, the connection of the mobile phone to the Internet is not permanent; it is only possible to connect to the Internet when a wireless network (cellular or WiFi) is available.

6. Variety: there are many mobile devices with different features; different CPU power, different screen sizes, different screen resolution, different Operating System, etc. It is hard to develop an application that works in all the different devices. This is the so-called the fragmentation problem, and is a challenge for the developer, who has to make an extra effort to ensure that the application will work in any device.

Examples of ICT tools for TC

Many tools for mobile devices are available to help disabled people in their communication (see Table 13-1). They all are Alternative and Augmentative Communication (AAC) tools, each one based on some multimedia elements. For example, JabTalkis based on real pictures. Most of the applications are based on pictograms. Arasaac is database of pictograms that are used in many AAC applications. Some applications allow the user to customize some pictograms, replacing them by a real picture taken with the mobile device (e.g., Words in Pictures and TalkInPictures). There are also applications whose main resource is a set of sign language video clips (e.g., Sign Language!). We can also find applications focused on providing information for caregivers, like Asperger's & Autism Community. However, as far as we know, there is no single application that contains information for caregivers and put together all the resources available for communication (pictograms, pictures and sign language video clips).

Table 13-1: Some AAC tools available for mobile devices.

Application	URL	Features
JabTalk	www.jabstone.com	Real pictures
AAC Speech Communicator	zemleris.com	Pictograms
Avaz	www.avazapp.com	Pictograms
CPA	www.comunicadorcpa.com	Pictograms
LetMeTalk	www.letmetalk.info	Pictograms
Pictotraductor	www.pictoaplicaciones.com	Pictograms

PiktoPlus Autism AAC	www.limbika.com/team/	Pictograms
Pixwriter	www.suncastletech.com	Pictograms
Proloquo2Go	www.assistiveware.com	Pictograms
Sc@ut	scaut.ugr.es/scaut/	Pictograms
Words in Pictures	www.fingertalks.it	Pictograms (can be replaced by real pictures)
Arasaac	arasaac.org	Database of pictograms
TalkInPictures	www.myautisticapps.com	Pictograms (can be replaced by real pictures)
Sign Language!	www.everydayasl.com	Sign language video clips
Asperger's & Autism Community	www.aspiescentral.com	Information for caregivers

The EC+ tool

In this section, we describe the EC+ tool developed in the context of an Erasmus+ KA2 project EC+-Enhancing Communication. The interested reader can find additional information of the project and the EC+ tool in the EC+ website[3].

This tool is composed of a mobile application to be installed in the smartphones or tablets and an academic portal that is available on-line. In practice, the mobile application should be the preferred way to access all these resources, since mobile devices are very common in our daily life. The main goal of these applications is to provide in a centralized location a valuable set of resources to ease the communication with disabled people having communication difficulties.

EC+ Requirements

In order to obtain this main goal, several requirements were defined for each application. The requirements of the mobile application are:

- The application must show a list of words relevant for a basic communication with disabled people.

[3] http://ecplusproject.uma.es

- Each word must have a series of multimedia resources associated with it that can help the user to communicate with disabled people.
- Three kinds of resource are considered: video clips with sign language interpretation of the word, pictograms with a clear image representing the word and pictures of real-world objects representing the word.
- The application must show a list of syndromes that could affect communication with a detailed description, potential treatment, and external references.
- The list of words and syndromes must be in four languages: Spanish, Catalan, Dutch, and German.
- Both lists (words and syndromes) must be downloaded and updated through Internet from an academic portal.
- The multimedia resources must be available in three different resolutions for them to be adaptable to the screen resolution and the network bandwidth of the mobile devices.

The requirements of the academic portal are:

- The academic portal must show to any user the list of words and syndromes (see the requirements of the mobile application above) and related multimedia resources in the three available resolutions and the four languages.
- Both lists (words and syndromes) and the multimedia resources must also be available through a Web Service (Alonso et al., 2010), where the mobile devices connect to download and update their content.
- Administrators of the academic portal must be able to log in with a user and password to edit the content of the academic portal.

The resources

The tool provides multimedia resources associated to a list of more than 400 words that are basic in any communication. These resources include (for each word):

- Video clips: The video clips, as the rest of multimedia resources, will be used to represent the different object and actions in order to ease the communication with disabled people having communication difficulties. In concrete, the clips show sign language interpretation of the word. In addition, we have created some video for some actions where pictures are not possible nor representative.

- Pictograms: A pictogram is a quite simple drawing schematizing the main features of the object or the action. Since they are created (usually by a specialized painter) instead of being taken from the real world, we can use lossless formats to represent it.
- Pictures: The picture of real-world objects representing the word in order to ease the understanding of some concepts.

All these resources were taken with the highest possible quality and, then, we apply some post-processing steps to correct small errors and to generate the final resources with the required format and resolution. The multimedia resources are available with three different qualities: low, medium and high quality. On the one hand, low and medium qualities are thought to be use in the mobile application to reduce the storage space and the bandwidth requirements (which are very important factor for this kind of devices). On the other hand, the higher qualities will be used in the web application in which the space and bandwidth limitations are not a problem. In any case, the user can decide which quality of resources used by his/her application.

This tool also contains information about 15 specific syndromes related to severe intellectual and developmental disabilities and special communications needs (e.g., Angelman, Pitt-Hopkins, sever autism, etc.).

These resources are available in four languages: Spanish, Catalan, Dutch, and German. In additional, the information about the syndromes is also available in English. As we said before, all the multimedia resources and the description of the syndromes have been originally created for this tool.

The Resulting Applications

The two applications of the EC+ tool are linked by mean of a Web Service (see Figure 13-2). The mobile application downloads the list of words, syndromes and resources from this web service, which is located in the same machine as the academic portal. Both, the web service and the academic portal access to exactly the same database and resource files, since they are run in the same machine.

Now, we will describe the two applications, we will start with the mobile one. The current mobile application works in devices with Android Operating System version 4.0.3 or higher. We chose Android because it is the most used smartphone Operating System (more than 85% of the mobile devices in the market run Android); and version 4.0.3 because than 99% of the Android devices use this or higher version, at

the time of writing these lines. This application can be directly installed from the Google Play (the Android Market).

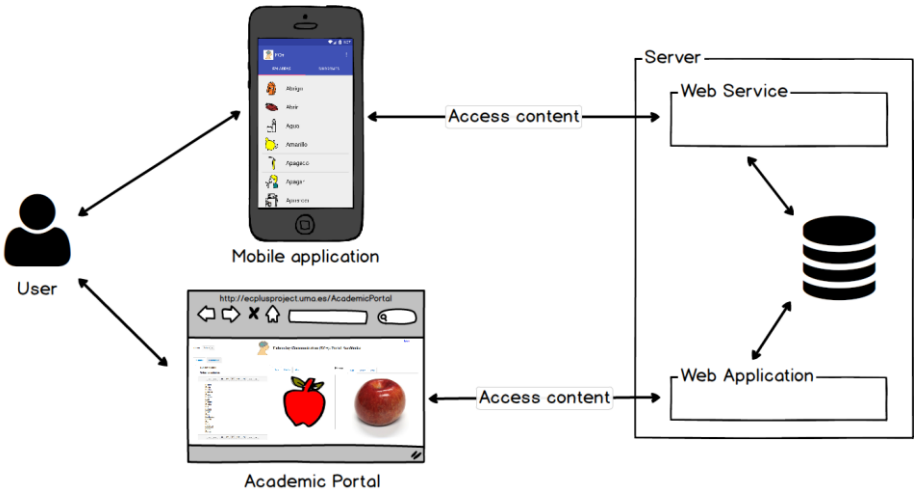

Figure 13-2: Architecture of the application.

The mobile application shows the list of words and syndromes in different panels (see Figure 13-3, left). In the case of the list of words, the pictogram of each word is also shown at the left of the word for a fast identification. If we click in a word we can access a new view where all the multimedia resources associated to that word (video clip, pictogram and picture) are shown (see Figure 13-3, center). The video clips can be reproduced and the pictogram and pictures can be enlarged. There are two lists of words: the basic ones and the advanced ones. The difference is related to the amount of available resources and the level of abstraction of the word itself. The set of advanced words contain, for example, all the prepositions, which are difficult to illustrate and require a higher level of abstraction.

The application also includes a panel with all the pictograms of the words (only the pictograms) and a list of general communication documents. The panel with pictograms can be used by the disabled people to point what s/he wants to communicate. The communication documents contain basic scientific information of a few concepts related to the communication, like the concept of total communication.

Each time the application is launched by the user, it contacts the web service to check if any of the resources or the lists have been updated. If there are new updates, they are downloaded. The mobile application downloads the resources in the language of the device, if there are resources in that language. However, the user can also select the language of the resources from the ones available. New languages and resources can

be added to the EC+ tool using the academic portal and they will be immediately available to be used in the mobile application.

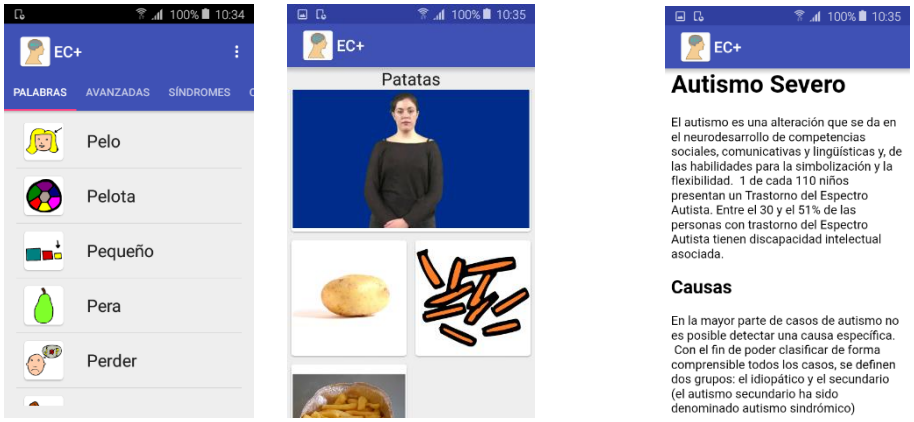

Figure 13-3: Screen capture of the list of words (left), resources of a word (center), and information of a syndrome (right) in the mobile application.

Currently, we are also developing a version of this application for iOS, which allows iPhone and iPad users to have access to the same information. With this version, we will cover most of the devices in the market: more than 99% of the mobile devices in the world.

The academic portal is an on-line website accessible using any web browser. The academic portal has two main uses. On the one hand, it allows accessing the resources (list of words and syndromes and multimedia elements for the words) in a similar way to the mobile application (see Figure 13-4). This is freely available for any user using the following URL: http://ecplusproject.uma.es/academicPortal/. On the other hand, it is also an administration tool to add new resources (languages, words, syndromes, and multimedia elements) or to update existing ones. These new or updated items will be immediately available for the rest of the users through the mobile application or the website. The ability of adding/updating items to the system will be restricted to some specific registered users approved by the administrator of the academic portal.

Figure 13-4: Screen capture of the academic portal showing the details of a word (left) and the description of a syndrome (right).

The future of ICT in total communication

Most of the applications that are available today to help in Total Communication are databases. They contain many resources to help in the communication with the persons having special communication needs. These resources can be videos, pictures, audios, symbols, etc. EC+ is not an exception; it contains a database of resources organized by words. Moreover, this is very useful in many cases.

However, taking into account the current advances in Computer Science,it is possible to apply also these advances to the field of total communication and conceive applications that do smarter things to help the disabled people.

One of the new advances of Computer Science that can be used is the Internet of Things. The idea of Internet of Things is to connect all the smart devices with Internet with the final goal of taking global smart decisions between the devices or control the network of things in some way. Smart sensors today are common and cheap. For example, there are many home automation solutions that allow the users to see what's happening at their home, open the windows or start the washing machine from a mobile phone. In the case of total communication, the Internet of things could be used to allow the disable people to communication not with a person, but with the things themselves, bypassing one communication step and improving their autonomy. For example, the disabled person could open the window or the door, by communicating with the window or the door directly.

The audio and video recognition can also be used in total communication. In this case, the disabled people simply talks, or make a gesture to a camera that is automatically understood by the devices, providing a direct communication with the machine.

Augmented reality adds virtual objects to a real scenario. This can be used to help the disable people by adding a hint in a scenario to ease the communication with

the environment. For example, it could be possible to add information to go to a room or to find the exit based on the image recorded by a mobile device.

Artificial Intelligence and Machine Learning are very popular domains nowadays. There are autonomous cars that are able to see the road and drive from an origin to a destination taking into account the other cars and pedestrians. There are computers that are winners in games like Chess or Go. And all this intelligence could also be used in total communication. For example, using machine learning techniques, the machine could learn how to better communicate with disabled people. This idea has already been applied in other contexts, like voice recognition. In the case of the voice recognition applications, there is a learning stage where the machines hear the voice of the user reading a given text. This learning idea can also be used in the communication with disabled people and the computer could learn the meaning of the different gestures, sounds and behaviors of the disabled user.

Conclusions

In this document, we have shown how ICTs can be useful tools for easing the communication with people with special communications needs. Specially, we have focused on mobile devices because they are portable devices that we can bring with us all the time. These devices have a set of advantages when they are used as tools in Total Communication, such as portability or versatility, but they also have some limitations, mainly related to their intrinsic nature (small screen or limited hardware resources).

We have also presented some ICT tools used to help disabled people in their communication. In particular, we have focus on the EC++ tool, a set of applications for easing the inclusion of people with severe intellectual or developmental disabilities in their communities. The tool allows to access the information and a large number of multimedia resources. These multimedia resources (videos, pictures and pictograms) were originally created for this tool in the context of an Erasmus+ KA2 project: EC+- Enhancing Communication. All this is available in four different languages (Spanish, Catalan, German and Dutch).

We also describe the two main components of our tool, the mobile application and academic portal, showing how they present the different resources. These resources can be added or updated with an authorized account through the academic portal.

Finally, we have discussed the utilization of some of the most recent advances in Computer Science (Artificial Intelligence, Machine Learning, Augmented Reality,

Internet of the Things…) in order to build applications that do smarter things to help the disabled people.

Acknowledgements

The work has been co- funded by the Erasmus+ program of the European Union (project 2015-1-ES01-KA203-015625), the excellence international campus Andalucía Tech and Samsung.

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4. EC+: App for People with Profound Intellectual Disability (PID)

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Augmentative and Alternative Communication (AAC) is defined as an area of clinical practice and research that seeks to compensate (temporarily or permanently) problems within the scope of expressive communication (i.e. severe damage in spoken or written production or in reading processes) or as an alternative to non-functional speech (NSW Government Clinical Guideline, 2016). Following the classic classification of Lloyd & Karlan (1984), which differentiates between AAC systems with or without aids.

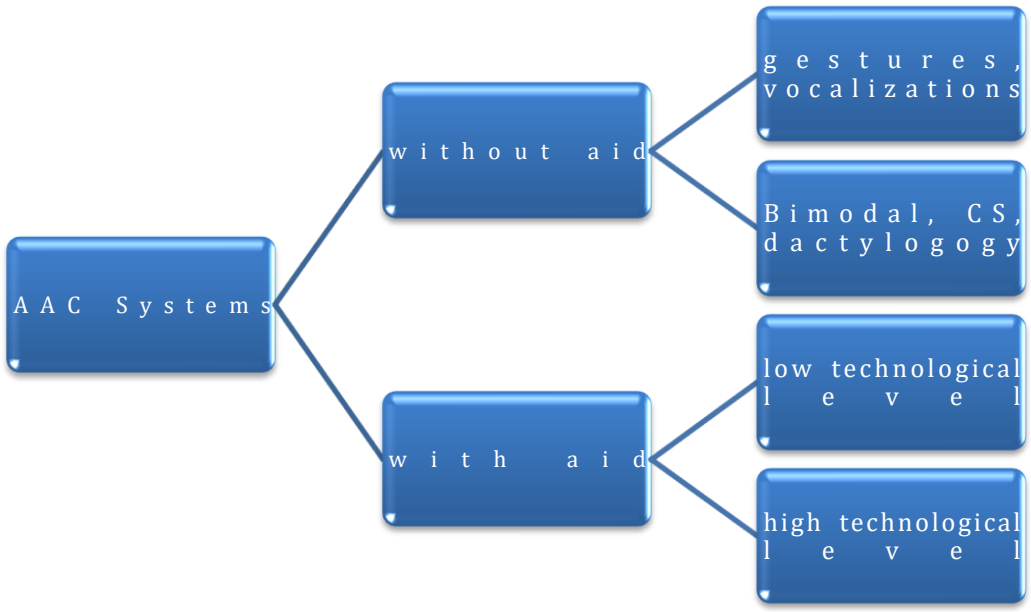

Figure1: by Marina Calleja

AAC Systems without aids (AAC-a) are those that are composed of forms of natural non-verbal communication (including gestures and facial expressions, in addition to signs). AAC-a systems (using their own body, not resorting to additional devices e.g. electronic equipment).

AAC systems with aids (AAC+a) are those that require the use of equipment, devices or additional material to the body itself, through which a person can communicate with the environment. The AAC +a refers to any element external to the subject itself such as, for example, object symbols, communication boards, cards, speech generating devices, computer, mobile phone, tablet... The AAC +a includes high and low technology systems.

The difference between AAC + high and low technological level is that the first are electronic devices and need energy to be used, while seconds do not.

Users of the AAC

Potential users of AAC are those who have serious difficulties communicating through speech throughout their lives (from childhood to adulthood). The causes of Complex Communication Needs (hereinafter CCN) may be physical, sensory, cognitive and environmental (for example, they may be caused by a hearing, cognitive or motor disability). Sometimes this limitation is permanent (e.g. in the case of some modalities of Cerebral Palsy and Intellectual Disability), while in other circumstances the limitation is temporary (e.g. after a surgical intervention on the vocal cords the patient cannot communicate through speech, but after the rehabilitation period, the patient can use speech again).

CCN usually refers to the fact that they cannot use speech as a means to communicate with other partners. However, in some cases, the subject can use speech (understood as vocal and verbal productions), although the degree of intelligibility achieved is so low that it fails to carry out functional communicative exchanges efficiently. In any of the cases cited, it is necessary to resort to strategies not based on the natural resource of speech, that is to say, in some AAC modality (Calleja & Rodríguez, 2018).

Within the AAC users group we find people with intellectual disability (ID). This group has been excluded for decades from intervention programs with AAC. The reason was that they did not meet the minimum cognitive prerequisites. But even those unintentional communicating subjects can be perceived as potential communicators since their behavior must be interpreted by the interlocutors (teachers, caregivers, assistant, healthcare staff, family…) as a communicative act (Brady et al, 2016).

ID is conceptualized as a disability characterized by significant limitations in both intellectual functioning and adaptive behavior, which encompasses many social skills and daily practices.

This disability originates before the age of 18 (Retrieved from http://aaidd.org/intellectual-disability/definition#.WpRKsHyCHIW) (Luckasson and Cols, 2011). The classification system has migrated from a model based on the deficit to another in which this disability is conceived as a situational construct, mediated by the communicative, sensory, motor and behavioral characteristics of a person as well as the

demands and supports associated to the contexts with which it interacts (Brady *et al*., 2016)

DSM-5 (APA, 2013) assumes the guidelines of the definition of the AAIDD and defines intellectual disability as "a disorder that begins during the period of development and that includes limitations of intellectual functioning as well as adaptive behavior in the conceptual, social and practical domains ".

ID is organized according to the DSM-5 depending on the adaptive functioning of the subject and we can distinguish between mild, moderate, severe and profound. Profound Intellectual Disabilities (PID)(some authors continue to use the term Severe Intellectual Disabilities) is present in the social area and entails a limited understanding of symbolic communication. Therefore, these people can understand simple gestures and generally make use of non-verbal communication (American Psychiatric Association, 2013).

When the presence of NCC is added to the PID, the clinical intervention becomes extremely complex unless it is used for Aumentative and / or Alternative Communication Systems (AAC) (Romski and Sevcik, 2005).

In the 1980s, subjects with PID were excluded as potential users of the AAC. As we mentioned before, the reason for this exclusion was because they either did not show the cognitive skills needed to correctly use the AAC Systems or they had some actual or potential speech skills, and it was thought that the use of a AAC System could interfere with a supposed potential development of speech to be achieve in the future with stimulation (National Joint Committee for the Communication Needs of Persons with Severe Disabilities, 2002; Snell, et al., 2003)

However, recent studies have shown that the use of one or several AAC systems not only does not reduce the development of speech skills (Millar, Light and Schlosser, 2006), but also the use of VOCA-type devices or the visual scenes, allow people with PID to increase their participation in class, at home and in social environments (Wilkinson and Henning, 2007); enable them to make choices (Cosbey and Johnston, 2006); help them improve their communication skills (Cheslock, Barton-Hulsey, Romski and Sevcik, 2008); and even, they contribute to modify the stereotypes and the perceptions that others have about them (Wilkinson and Henning, 2007).

Lund, Quach, Weissling, McKelvey & Dietz (2017) suggest that before proposing a AAC device we would have to carry out: a) an evaluation centered on the

individual with CCN, b) an assessment of the skills of the communication partners and c) an analysis of the characteristics of the device.

In relation to the characteristics of the individuals, some information about their communicative skills must be available for researchers or speech language therapists. These professionals must be aware of which are their needs and their communication objectives; assess the strengths of the communicative ability of the individual, the barriers that can limit the communicative exchange as well as the preferences. For the specific case of subjects with ID, decision making about the AAC is made according to the subjects' needs. In this sense, Reichle, York, York-Barr & Sigafoos (1991) proposed that we must pay attention to the way in which the subject carries out communicative exchanges so we must know which are the vocabulary needs to meet their communication needs according to their activities and routines, we must know which communicative attempts the subject should learn. We would even have to assess whether the best option is an strategy with or without aid. Given the possibility of choosing the option of AAC without aid, we would have to assess the user's motor skills as well as the type of vocabulary he or she needs to learn. While in the modality of AAC with aid (AAC+a), it would be necessary to evaluate the type of symbol (photograph vs. drawing), the size of the symbol, the representation of the message (nature and length), the way to access to the message (through direct selection, scanning or eye movements), the response options (visual presentation or synthesized speech), the portability of the device and the ability to initiate episodes of social interaction (ability to call attention to a communication partner or the possibility of starting communicative exchanges) (van der Meer et al., 2011)

In relation to the skills of the interlocutors (professional team, staff, family, ...) that are going to communicate with the PID, it is important to know if they have had previous experience with the use of AAC devices, to know their acceptance or rejection attitude for the use the AAC devices. Even Thistle & Wilkinson (2013) pointed out that sometimes before opting for a device, clinicians have to add vocabulary related to the preferences of family members or the courtesy formulas they usually use.

Finally, when opting for an AAC system, we know that this system can take many forms including gestures and manual signs of sign language, visual symbols inserted in boards or communication books, written words, drawings or electronic devices that provide vocal answers (Beukelman & Mirenda, 2015). Among the AAC

systems with electronic assistance are computers, artificial speech generation devices or mobile technologies with applications for communication (Thistle & Wilkinson, 2013).

EC+ APP

EC + (Chicano, Postigo, Luque & Calleja, 2018) is an app for tablets and mobile phones for IOS and Android developed at the University of Malaga for people with DIP and the interlocutors of their environment, whose purpose is to increase communication exchanges. The mobile application is based on the API (Application Programming Interface) in version 4.0.3. of Android (Ice Cream Sandwich) so that it can be used in most of the existing mobile devices in the market.

The acronym EC+ comes from the full English name of the Erasmus + project where this app has been developed, *Enhancing Communication or Improving Communication*. Its purpose is to serve as an AAC device of high technological level based on the assumptions of the multi-modality (simultaneous presentation of visual, gestural and acoustic stimuli such as pictograms, hand signs, photographs and words-spoken and written-).

The approach of EC+ is multimodal. When you access to the interface, to select a certain symbol it is enough to click on the desired icon and the symbol appears in five different modalities (manual sign with linguistic value, verbal-vocal production, written presentation, pictographic representation with drawing and pictographic representation with photographs and video). The process to go from one screen to another can be done by sliding one's finger.

In addition, the app has a series of medical and intervention guides for diseases and syndromes that deal with PID + NCC, as well as instructions to carry out communicative exchanges.

The contents of the app are presented in five different languages (Spanish, English, German, Dutch and Catalan).

Where does EC+ arise?

The app has been developed from the University of Malaga, within the framework of a European Project within the Erasmus Plus program called Enhancing Communication: Research to Improve Communication for People with Special Needs and Development of ICT Resources and Tools. It is funded by the European Union and involves various academic institutions (University of Malaga, the University of Ghent

(Belgium), the University of Klagenfurt (Austria) and a specialized center Parc Taulí Hospital (Sabadell).

Aim

The purpose of the app is to serve as an AAC device with a high technological level based on the assumptions of multimodal communication (pictograms, manual signs, photographs and words) for people with PID+ CCN as well as caregivers and their relatives. Also the app was conceived as an instrument to reduce anxiety on the part of professionals from different fields (healthcare, education, interpreting, leisure…) when they have to interact with PID and CCN people and they have not previous specialized knowledge of intellectual disability.

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5. Base de datos terminológica sobre un trastorno del lenguaje: la afasia

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Las dificultades en comunicación social, concretamente en contextos sanitarios, se agravan cuando se ven involucradas varias lenguas. Si bien en los países europeos la normativa prevé que todos los ciudadanos tengan acceso a los servicios sanitarios de manera igualitaria, esto solo puede garantizarse si los servicios de traducción y, sobre todo, de interpretación están convenientemente preparados para cubrir la demanda. Esto no solo se consigue con un número suficiente de profesionales con formación adecuada: cada vez hay más publicaciones que hacen hincapié en la importancia del dominio de la terminología específica por parte del intérprete así como en que sepa emplear los recursos y herramientas para la gestión terminológica.

Para el proceso de traducción ya existen desde hace tiempo trabajos sobre el porcentaje que supone la búsqueda terminológica sobre el total del tiempo empleado por el traductor, y resulta que llega a ser hasta un 75 %. Para la interpretación aún no disponemos de tales estudios, aunque son cada vez más los trabajos que hacen referencia a cómo simplificar la preparación terminológica del encargo de interpretación para ahorrar tiempo, pues existe la conciencia que esta parte –por estar estrechamente ligada al trabajo conceptual– resulta crucial para el intérprete. El motivo reside en que la mayoría de los discursos especializados expresa un porcentaje elevado de conceptos específicos, reflejados en términos. Por eso es fundamental que el intérprete se informe bien sobre la nomenclatura del campo de conocimiento al que pertenece el discurso que interpretará y, a continuación, rastree y filtre bien los términos correspondientes a los conceptos que conforman la estructura de ese ámbito especializado. Así, cumplirá con dos objetivos: llegará a un mayor entendimiento del tema en cuestión por leer necesariamente textos sobre la materia, y además llegará a conocer los términos que pueden surgir durante la interpretación. Además, una buena preparación terminológica ayudará a mantener la consistencia en el uso de la terminología.

Esta base de datos se centra en uno de los trastornos del lenguaje más frecuentes: la afasia. Se creó a partir de un corpus comparable trilingüe (español peninsular – inglés

americano – alemán estándar), compilado mediante el programa Sketch Engine. Con la ayuda de sus herramientas, se elaboró una lista de candidatos a término y se eliminaron las unidades que no pertenecían estrictamente al ámbito estudiado.

La presente base de datos se puede ordenar de manera bilingüe o trilingüe en cualquiera de las direcciones de las tres lenguas consideradas. Asimismo, se puede consultar como glosario simple (*Dictionary*) por orden alfabético bilingüe o trilingüe, con indicación de un contexto (haciendo clic en el enlace del término) y la definición del concepto (*Dictionary -> Concept -> View*).

Concept ID (termbases)	Sketch Engine	Fuentes/enlaces
1004253517 educación inclusiva	EN file5637300 ES file5637736 DE file5638720	- LD@home https://www.ldathome.ca/what-are-lds/glossary/ - Gesellschaft für Aphasieforschung und -behandlung (GAB) und Deutsche Gesellschaft für Neurotraumatologie und Klinische Neuropsychologie (DGNKN) http://www.aphasiegesellschaft.de/index.php/download_file/view/109/87/ - UNICEF https://www.unicef.org/mexico/spanish/mx_19I-La_inclusion_de_NN_con_transtorno.pdf
1006208275 cáncer	EN file5637090 ES file5637765 DE file5638605	- DKMIC https://www.dkmic.de/patienten/forum/chemotherapie/ - MULTIPLE MYELOMA. Research Foundation https://www.themmrf.org/multiple-myeloma-knowledge-center/glossary/ - CDC. Centros para el Control y la Prevención de Enfermedades https://www.cdc.gov/spanish/cancer/survivorship/resources/glossary.htm
1009233721 comunicación	EN file5637407 ES file5637574 DE file5638712	- CogniFithttps://blog.cognifit.com/es/afasia-de-broca/ - Wikipedia https://de.wikipedia.org/wiki/Apallisches_Syndrom - Wikipedia https://en.wikipedia.org/wiki/Broca%E2%80%99s_aphasia
1012567393 deterioro lingüístico	EN file5637029 ES file5637574 DE file5638567	- Revista Debates IESA https://pide.files.wordpress.com/2007/06/fco-javier-perez-deterioro-del-leng.doc - CogniFit https://blog.cognifit.com/es/afasia-de-broca/ - University of Cologne https://www.hf.uni-koeln.de/data/penke/File/Penke_syntactic%20disorders(1).pdf - Beobachter Gesundheit https://www.beobachter.ch/gesundheit/krankheit/aphasie-sprachstorung
1018431162 agramatismo	EN file5637029 ES file5637729 DE file5638812	- Estefalogoflipe's Blog https://estefalogoflipe.wordpress.com/2010/01/08/glosario-logopedico/ - iinnuar https://cordobapsicopedagogianeuropsicologia.com/2016/03/08/afasia-tipos-y-caracteristicas/ - Kliniken Schmieder http://www.kliniken-schmieder.de/behandlungs-spektrum/neurologische-erkrankungen/aphasie.html - University of Cologne https://www.hf.uni-koeln.de/data/penke/File/Penke_syntactic%20disorders(1).pdf
1019047183afásico	EN	- NEUROfest http://www.untitledtheater.com/glossary.html

	file5637114 ES file5638058 DE file5638798	- Amarysm https://amarysm.wordpress.com/trastornos-del-lenguaje/ - Wie vom Blitz getroffen http://www.logopaedie-oerlinghausen.de/wir-behandeln/sprache/aphasie/
1024242791 semántico	EN file5637128 ES file5638058 DE file5638731	- The Free Dictionary https://medical-dictionary.thefreedictionary.com/dysphasia - Gabinete Logopédico y Pedagógico http://www.logopedia-granada.com/afasia.htm - Hausarbeiten.de http://www.hausarbeiten.de/faecher/vorschau/62039.html
1033928276 admisión hospitalaria	EN file5637015 ES file5637616 DE file5638433	- INSALUD www.sedom.es/wp-content/themes/sedom/documentos/guia_sadc_definitiva.pdf - Celgene https://www.diagnose-psoriasis.de/service/glossar/ - Hospital Regional Alta Especialidad http://hraei.net/doc/2014/procedimientos-dir-operaciones.pdf
1034287147 atención ambulatoria	EN file5637524 ES file5637616 DE file5638583	- Hospital Regional Alta Especialidad http://hraei.net/doc/2014/procedimientos-dir-operaciones.pdf - NOVAFON https://www.novafon.com/de/news/aphasie-bei-kindern-und-jugendlichen - Hospital Regional Alta Especialidad http://hraei.net/doc/2014/procedimientos-dir-operaciones.pdf - TreatmentSolutionshttps://www.treatmentsolutions.com/dictionary/
1059076166 lóbulo	EN file5637070 ES file5637774 DE file5638595	- Motor Apraxia and Joubert http://diaridunaapraxia.blogspot.com/2011/02/definiciones-agnosia-afasia-disfasia.html - AllergieTEST http://allergietest.online/tierhaarallergie/ - Traumatic Brain Injury (TBI) https://tbi.cemmlibrary.org/Resources/Glossary
1059503877 perseveración afásica	EN file5637289 ES	- SCRIBD https://es.scribd.com/doc/286174873/manual-de-afasia-y-terapia-de-la-afasia - SCRIBD https://es.scribd.com/doc/286174873/manual-de-afasia-y-terapia-de-la-afasia - Indiana Speech-Language-Hearing Association

	file5637677 DE file5638411	http://www.islha.org/Resources/Documents/ODellConcepts%20Aphasia%20ISLHA%20B_W.pdf - Facharzt für Neurologie, Psychiatrie und Facharzt für Psychotherapeutische Medizin, Psychoanalyse - http://www.neuro24.de/show_glossar.php?id=1248
1075010886 intelectual	EN file5637014 ES file5637630 DE file5638677	- dbl Deutscher Bundesverband für Logopädie e.V. https://www.dbl-ev.de/kommunikation-sprache-sprechen-stimme-schlucken/stoerungen-bei-kindern/stoerungsbereiche/sprache/kindliche-aphasie.html - Rincón de Logopedia http://rincondelogopedia.blogspot.com/2009/07/glosario-de-terminos-de-logopedia.html - SCRIBD https://www.scribd.com/document/145619420/Speech-Glossary
1087766768 déficit	EN file5637389 ES file5638058 DE file5638764	- Patient. Making lives better https://patient.info/doctor/dysarthria-and-dysphasia - Altenpflege Ausbildung https://www.altenpflegeschueler.de/krankheiten/aphasie/ - Amarysmhttps://amarysm.wordpress.com/trastornos-del-lenguaje/
1089087450ansiedad	EN file5637524 ES file5637669 DE file5638799	- Conoce el autismo http://conoceelautismo.blogspot.com/2013/04/glosario-de-terminos-de-educacion.html - Treatment Solutions https://www.treatmentsolutions.com/dictionary/ - Gabinete de Psicología y Salud de Montse Valls Giner http://www.montsevalls.com/es/glosario01.htm - HaltenRaum http://haltenraum.com/article/expressive-aphasie
1095107991 afasia global	EN file5637545 ES file5637729 DE file5638816	- Facebook https://www.facebook.com/note.php?note_id=224336604250232 - Córdoba Pedagogía Neuropsicología https://cordobapsicopedagogianeuropsicologia.com/2016/03/08/afasia-tipos-y-caracteristicas/ - Identify the Signs of Communication Disorder https://identifythesigns.org/signs-of-speech-and-language-disorders/ - VITAES https://www.vitaes.de/aphasie/
1095421287 conocimiento	EN file5637250 ES file5637568 DE file5638573	- Children'sNeuropsychologicalServices http://www.childrensneuropsych.com/parents-guide/glossary/ - Fach Verbaende https://www.fachverbaende.net/vfp/no-jos/pruef/pruef_arb/27.pdf - Planahttps://planana.wordpress.com/2012/01/01/glosario-de-termino-de-voz-habla-y-lenguaje/

1096873745 conciencia	EN file5637389 ES file5637614 DE file5638812	- Kliniken Schmieder http://www.kliniken-schmieder.de/behandlungs-spektrum/neurologische-erkrankungen/aphasie.html - Sv Neurología http://www.svneurologia.org/libro%20ictus%20capitulos/glosario.pdf - Patient. Making lives better https://patient.info/doctor/dysarthria-and-dysphasia
1099414150disfunción	EN file5637443 ES file5637652 DE file5638704	- Fonomass http://fonomass.blogspot.com/2010/12/glosario-tecnico-achaf-asociacion.html - Judy Duchan. A History of Speech- Language Pathology http://www.acsu.buffalo.edu/~duchan/dictionary.html - Kinder & Jugenärzte im Netz https://www.kinderaerzte-im-netz.de/krankheiten/epilepsie/glossar/
1102196346 anomalía	EN file5637032 ES file5637630 DE file5638537	- Right Diagnosis from healthgrades http://www.rightdiagnosis.com/symptoms/eye_symptoms/glossary.htm - Estefanialogos https://estefanialogos.wordpress.com/glosario-logopedia/ - Symptomms http://symptomms.com/de/pages/1583847
1106508320 neuropsicología	EN file5637211 ES file5637647 DE file5638526	- Conoce el autismo http://conoceelautismo.blogspot.com/2013/04/glosario-de-terminos-de-educacion.html - SCRIBD https://www.scribd.com/document/62805309/Turnbull-Solms-Glossary - HaltenRaum http://haltenraum.com/article/aphasie - Pon un logopeda en tu vida https://anaagcaro.wordpress.com/glosario-logopedico/
1106892101 lóbulo temporal	EN file5637021 ES file5637677 DE file5638772	- CogniFit https://www.cognifit.com/es/partes-del-cerebro - Right Diagnosis from healthgrades http://www.rightdiagnosis.com/symptoms/asymmetrical_face/glossary.htm - SCRIBD https://es.scribd.com/doc/286174873/manual-de-afasia-y-terapia-de-la-afasia - Onmeda.dehttp://www.onmeda.de/krankheiten/aphasie-symptome-wernicke-aphasie-3689-5.html
1118595296 apraxia	EN file5637470 ES	- Planana https://planana.wordpress.com/2012/01/01/glosario-de-termino-de-voz-habla-y-lenguaje/ - Motor Apraaxia and Joubert http://diaridunaapraxia.blogspot.com/2011/02/definiciones-agnosia-afasia-disfasia.html

	file5637774 DE file5638831	- NetDoktor https://www.netdoktor.de/symptome/apraxie/ - Aphasiatoolbox.com https://aphasiatoolbox.com/glossary/
1118676708daño	EN file5637177 ES file5637673 DE file5638526	- HaltenRaum http://haltenraum.com/article/aphasie - Periodico de Saludhttp://periodicosalud.com/afasia-definicion-causas-tipos-sintomas-tratamiento/ - Medlineplushttps://medlineplus.gov/magazine/issues/spring17/articles/spring17pg9.html
1129228613afasia	EN file5637013 ES file5637688 DE file5638820	-
1130424084epilepsia	EN file5637193 ES file5637677 DE file5638414	- SV Neurología http://www.svneurologia.org/libro%20ictus%20capitulos/glosario.pdf - SCRIBD https://es.scribd.com/doc/286174873/manual-de-afasia-y-terapia-de-la-afasia - Carmen Kuchinke Logopädie http://www.logopaedie-kuchinke.de/glossar/buchstabe/a - Right Diagnosis form healthgradeshttp://www.rightdiagnosis.com/symptoms/eating_symptoms/glossary.htm
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1990514253 Wernicke	EN file5637407 ES file5637688 DE file5638812	- Historia de la Medicina www.historiadelamedicina.org/wernicke.html - Wikipedia https://en.wikipedia.org/wiki/Broca%E2%80%99s_aphasia - Kliniken Schmieder. Neurologisches Fach- und Rehabilitationskrankenhaus http://www.kliniken-schmieder.de/behandlungs-spektrum/neurologische-erkrankungen/aphasie.html - Facebook https://www.facebook.com/note.php?note_id=224336604250232
2006383736foniatría	EN file5639725 ES file5637602 DE file5638770	- INR LGII http://iso9001.inr.gob.mx/Descargas/iso/doc/MG-SAF-35.pdf - Pvb Praxisverbund Berlin. Prof. Dr. O. König http://www.hno-berlin-mitte.com/schwerpunkte/stimmstoerung-berlin/ - SciELOhttp://www.scielo.br/scielo.php?script=sci_arttext&pid=S1808-86942015000600581
2007602179cognitivo	EN file5637443 ES file5638058 DE	- Judy Duchan. A History of Speech-Language Pathology http://www.acsu.buffalo.edu/~duchan/dictionary.html - Gabinete Logopédico y Pedagógico http://www.logopedia-granada.com/afasia.htm - HaltenRaum http://haltenraum.com/article/aphasie

	file5638526	
2019083155 ambulatorio	EN file5637050 ES file5637616 DE file5638583	- WebMD https://www.webmd.com/arthritis/septic-arthritis-symptoms-diagnosis-and-treatment - NOVAFON https://www.novafon.com/de/news/aphasie-bei-kindern-und-jugendlichen - Hospital Regional Alta Especialidad http://hraei.net/doc/2014/procedimientos-dir-operaciones.pdf
2027823424 logopedia	EN file5637024 ES file5637647 DE file5638742	- My Handicap. My Chance https://www.myhandicap.de/gesundheit/koerperliche-behinderung/aphasie/ - Talking Point http://www.talkingpoint.org.uk/sites/talkingpoint.org.uk/files/afasicglossary13.pdf - Gabinete Logopédico y Pedagógico http://www.logopedia-granada.com/diccionario.htm
2041980303 comprensión auditiva	EN file5637104 ES file5637677 DE file5638548	- CVC Cervantes https://cvc.cervantes.es/ensenanza/biblioteca_ele/diccio.../comprensionauditiva.htm - SCRIBD https://es.scribd.com/doc/286174873/manual-de-afasia-y-terapia-de-la-afasia - Neupsy Key https://neupsykey.com/language-and-speech-disorders-aphasia-and-aphasic-syndromes/ - GFMK Ratgeber. Schlaganfall & Aphasie http://daniels-innovation-gesundheit.de/data/documents/Ratgeber_Aphasie_Schlaganfall_2014_low_web-1.pdf
2058274105 parafasia	EN file5637029 ES file5637729 DE file5638840	- Prezi https://prezi.com/2o1rbtciobsx/afasia-disfasia/ - Iinnuar https://cordobapsicopedagogianeuropsicologia.com/2016/03/08/afasia-tipos-y-caracteristicas/ - http://vonderheide.me/aphasie/aphasie.htm - University of Cologne https://www.hf.uni-koeln.de/data/penke/File/Penke_syntactic%20disorders(1).pdf
2059437854audición	EN file5637203 ES file5637734 DE file5638828	- Unbeschwert Hören https://unbeschwert-hoeren.de/hoeren/hoersturz/ - LUCIALOGOPEDIA https://lucialogopedia.wordpress.com/definiciones-relacionadas-con-logopedia/ - ENCYCLOPEDIA.COM http://www.encyclopedia.com/psychology/encyclopedias-almanacs-transcripts-and-maps/expressive-language-disorder
2066754612identificar	EN file5637493	- medikamente-per-klick.de https://www.medikamente-per-klick.de/apotheke/krankheitenglossar/borreliose/

	ES file5637674 DE file5638679	- Drake Institute of Behavioral Medicine http://www.drakeinstitute.com/auditory-language-disorder-treatment - Concept of Physio https://concept-of-physio.weebly.com/glosario-de-fisioterapia
2071056321concepto	EN file5637443 ES file5637704 DE file5638712	- Judy Duchan. A History of Speech-Language Pathology http://www.acsu.buffalo.edu/~duchan/dictionary.html - Conoce el autismo http://conoceelautismo.blogspot.com/2013/04/glosario-de-terminos-de-educacion.html - Wikipedia https://de.wikipedia.org/wiki/Apallisches_Syndrom
2074423330 neuroimagen	EN file5637554 ES file5637677 DE file5638796	- neuroimagen - enciclopediasalud.com www.enciclopediasalud.com/definiciones/neuroimagen - Mein Leben mit NET https://www.mein-leben-mit-net.de/glossar/ - Advances in Language and Literary Studies http://files.eric.ed.gov/fulltext/EJ1128489.pdf - SCRIBD https://es.scribd.com/doc/286174873/manual-de-afasia-y-terapia-de-la-afasia
2110060821 mutismo	EN file5637278 ES file5637688 DE file5638583	- ACV- Reflexiones http://acv-reflexiones.blogspot.com/2008/08/afasia-causas-clasificacion-y.html - NOVAFON https://www.novafon.com/de/news/aphasie-bei-kindern-und-jugendlichen - Right Diagnosis from healthgradeshttp://www.rightdiagnosis.com/a/aphasia/glossary.htm
2110097682parte del cuerpo	EN file5637349 ES file5638026 DE file5638759	- Lewy Body Dementia Association https://www.lbda.org/downloads/LBDGlossary.pdf - Deutsche Hochdruckliga e.V. DHL https://www.hochdruckliga.de/glossar.html - GenesisCarehttps://www.genesiscare.es/glosario/
2114862478capacidad	EN file5637466 ES file5637688 DE	- Bel Marra Health. Doctor Formulated Supplements https://www.belmarrahealth.com/aphasia-vs-dysphasia-differentiating-symptoms-causes/ - Medizin. Alle über Gesundheit und gesunde Lebensstile http://omedicine.info/de/alexic-anomia.html - ACV- Reflexiones http://acv-reflexiones.blogspot.com/2008/08/afasia-causas-clasificacion-y.html

	file5638718	
2115058863 comprensión	EN file5637177 ES file5637688 DE file5638663	- Onmeda.de http://www.onmeda.de/krankheiten/aphasie-diagnose-3689-6.html - NIH National Institute on Deafness and Other Communication Disorders https://www.nidcd.nih.gov/sites/default/files/Documents/health/voice/Aphasia6-1-16.pdf - ACV- Reflexiones http://acv-reflexiones.blogspot.com/2008/08/afasia-causas-clasificacion-y.html
2135140662 transcortical	EN file5637128 ES file5637677 DE file5638829	- SCRIBD https://es.scribd.com/doc/286174873/manual-de-afasia-y-terapia-de-la-afasia - SCRIBD https://es.scribd.com/doc/286174873/manual-de-afasia-y-terapia-de-la-afasia - HaltenRaum http://haltenraum.com/article/aphasie - The Free Dictionary https://medical-dictionary.thefreedictionary.com/dysphasia
2136198072responder	EN file5637130 ES file5637669 DE file5638844	- curendo https://www.curendo.de/pflege/demenz-sprachfaehigkeit-ueberpruefen/ - AUTISM SPEAKS https://www.autismspeaks.org/sites/default/files/docs/glossary_and_resources_0.pdf - Gabinete de Psicología y Salud de Montse Valls Giner http://www.montsevalls.com/es/glosario01.htm
2141854377 conversación	EN file5637470 ES file5637618 DE file5638764	- SCRIBD https://www.scribd.com/document/296108932/Afasia-tratamiento - Altenpflege Ausbildung https://www.altenpflegeschueler.de/krankheiten/aphasie/ - Aphasiatoolbox https://aphasiatoolbox.com/glossary/
2146803339memoria	EN file5637090 ES file5637618 DE file5638416	- MEDICLIN. MediClin Klinikum Soltau http://www.mediclin.de/Portaldata/24/Resources/pdf_cp/zielgruppe_patienten/glossar/aphasie-station-soltau.pdf - SCRIBD https://www.scribd.com/document/296108932/Afasia-tratamiento - Multiple Myeloma Research Foundation https://www.themmrf.org/multiple-myeloma-knowledge-center/glossary/

6. Glossaries for interpreters: 15 syndromes described in EC+ (English/Spanish/German)

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Interpreting in healthcare settings has always been a risky practice and a real challenge for linguistic professionals, especially when it comes to the field of rare diseases. These diseases, which the European Union defines as those that affect less than 5 people out of every 10,000 in the general population, are usually pushed aside in favour of more common diseases, so an interpreter may think twice before deciding to interpret for a family member of a patient who suffers from one of these illnesses, or may have problems when interpreting in medical congresses. In these cases, not only is there the obstacle of having to interpret from one language into another, but there is also the added problem of specialised terminology, this being a discipline that provides a name to different concepts that belong to a specialised subject field (Bowker, 2009, p.286). However, and in spite of the problems mentioned, medical interpreters are key factors in communication in the healthcare field, as they make communication easier between patients, doctors and other healthcare providers, especially if the latter do not have a high knowledge of foreign languages (Varela Salinas and Meyer, 2015, p.7).

Medical interpreting has been developing very slowly over the last 50 years (Roat and Crezee, 2015, p.238). Furthermore, according to Postigo Pinazo (2015, p. 53) a high percentage of interpreting students are quite wary of interpreting in healthcare settings in risky situations. This could be due to the fact that practice in healthcare discourse and face-to-face bilateral interpreting is not very common in the training of translation and interpreting students. Moreover, as stated by a survey carried out on students by Postigo Pinazo (2015), as little as 20 % of the subjects had a habit of compiling biosanitary and medical-related glossaries. All the mentioned factors have had a negative impact on medical interpreting, and an even bigger one on communication involving rare-disease matters. Having become aware of the above-mentioned handicaps, one of the EC+ Enhancing Communication project's objectives is to provide interpreters with specialised documentary tools and resources, such as specialised glossaries, in order to provide a high-quality interpreting service.

Due to the lack of available glossaries relating to rare diseases, especially in any language that is not English, a trilingual glossary, compiled using three languages from the consortium (English, Spanish and German), is being presented here. There are 15 glossaries in total, one for each of the rare diseases, and the terms are mainly related to the description, interventions, medical indications, treatments and symptoms of these

illnesses. These 15 syndromes were selected by Parc Taulí Hospital in Sabadell because they seriously affect communication; in other words, the individuals that are affected by these diseases find it hard to communicate with other people and vice versa.

The selected terms from the glossaries were extracted from the documents related to the diseases in the EC+ project web page, from abstracts , association web pages (for example the Angelman Syndrome Foundation and the Spanish *Asociación del Síndrome de Angelman*) and from Orphanet (amongst other sources), which is a Spanish database for rare diseases. Orphanet is one of the very few databases that exist relating to this field and is reliable as it has been created by scientists. The only downfall is that its data collection is quite small and is only in the Spanish language. So, once again, it is crucial to highlight that these trilingual glossaries represent an innovative project based on real needs, as the few databases and glossaries that are available up to date are usually monolingual. Some of these terms were either selected manually, normally based on how frequently they are used, or put through the concordance programme AntConc which, besides showing words from a text in their context, also shows those words which appear most frequently in a set of texts. This process was used for extracting the terms in every single one of the 15 rare diseases that are the focus of this project. The equivalents of the words in other languages were found by comparing parallel texts from specialised and informative abstracts, scientific journals and specialised journals.

In conclusion, there is a niche to be filled with regard to terminology in the field of rare diseases, and interpreters need terminology resources such as electronic multilingual glossaries to be able to transmit important information and avoid the consequences of miscommunication in healthcare scenarios as much as possible. These glossaries, which will be developed and completed further on, offer new resources for interpreters.

The 15 different glossaries in English, Spanish and German are presented here below, listed in alphabetical order of the chosen syndromes: Angelman syndrome, cerebral palsy (CP) in children, dystonic tetraparesis, Cornelia de Lange syndrome, deafness associated with other causes of intellectual disability, infantile epileptic encephalopathies: Dravet syndrome, Kleefstra syndrome, Lennox Gastaut syndrome, MECP2: Rett syndrome, microdelation and microduplication syndromes with serious language affectation, Mowat-Wilson syndrome, Phelan-McDermid syndrome, Pitt-Hopkins syndrome, severe autism, Usher syndrome and West's syndrome.

1. Angelman syndrome

ENGLISH	**SPANISH**	**GERMAN**	**REFERENCES**
Angelman's	Síndrome de	Angelman-Syndrom	http://www.medigraphic.co

syndrome	Angelman		m/pdfs/abc/bc-2002/bc023h.pdf
Atypical swallowing	Deglución atípica	atypisches Schlucken	http://www.coem.org.es/sites/default/files/revista/cientifica/vol7-n1/61-64.pdf
Balance problem	Problema de equilibrio	Gleichgewichtsstörung	http://www.medigraphic.com/pdfs/abc/bc-2002/bc023h.pdf
Bands 11-13	Bandas11-13	Chromosomenbänder 11-13	http://www.medigraphic.com/pdfs/abc/bc-2002/bc023h.pdf
Behavioural phenotype	Fenotipo conductual	Verhaltensphänotyp	http://www.analesdepediatria.org/es-pdf-S1695403308720526
Cerebral palsy	Parálisis cerebral	Zerebralparese	http://www.analesdepediatria.org/es-pdf-S1695403308720526
Chewing problems	Problemas de masticación	Kaubeschwerden	http://www.coem.org.es/sites/default/files/revista/cientifica/vol7-n1/61-64.pdf
Chromosomic alteration	Alteración cromosómica	chromosomale Veränderung	http://www.medigraphic.com/pdfs/abc/bc-2002/bc023h.pdf
Clinical criteria	Criterio clínico	klinische Beurteiling	http://www.analesdepediatria.org/es-pdf-S1695403308720526
Deletion	Deleción	Deletion	http://www.medigraphic.com/pdfs/abc/bc-2002/bc023h.pdf
Dental literature	Literatura dental	zahnmedizinische Literatur	http://www.coem.org.es/sites/default/files/revista/cientifica/vol7-n1/61-64.pdf
Developmental delay	Retraso psicomotor	verzögerte psychomotorische Entwicklung	http://www.medigraphic.com/pdfs/abc/bc-2002/bc023h.pdf
Diastemas	Diastemas	Diastema	http://www.coem.org.es/sites/default/files/revista/cientifica/vol7-n1/61-64.pdf

Dismorphic feature	Característica dismórfica	dysmorphische Merkmale	http://www.coem.org.es/sites/default/files/revista/cientifica/vol7-n1/61-64.pdf
Drooling	Babeo	vermehrter Speichelfluss	http://www.coem.org.es/sites/default/files/revista/cientifica/vol7-n1/61-64.pdf
Epilepsy	Crisis convulsiva	Krampfanfall	http://www.medigraphic.com/pdfs/abc/bc-2002/bc023h.pdf
Facial dysmorphism	Dismorfia facial	faziale Dysmorphie	http://www.analesdepediatria.org/es-pdf-S1695403308720526
Fluorescence *in situ* hybridization	Hibridación fluorescente *in situ*	fluoreszierende In-Situ Hybridisierung (FISH)	http://www.medigraphic.com/pdfs/abc/bc-2002/bc023h.pdf
Fluttering movement	Movimiento de aleteo	Flatterbewegungen	http://www.medigraphic.com/pdfs/abc/bc-2002/bc023h.pdf
Frequent smiling	Sonrisa constante	fortwährendes Lächeln	http://www.medigraphic.com/pdfs/abc/bc-2002/bc023h.pdf
Genetic counseling	Consejo genético	genetische Beratung	http://www.medigraphic.com/pdfs/abc/bc-2002/bc023h.pdf
Genetic disease	Enfermedad genética	genetisch bedingte Krankheit	http://www.medigraphic.com/pdfs/abc/bc-2002/bc023h.pdf
Hyperactivity	Hiperactividad	Hyperaktivität	http://www.medigraphic.com/pdfs/abc/bc-2002/bc023h.pdf
Hypotonia	Hipotonía	Hypotonie	http://www.medigraphic.com/pdfs/abc/bc-2002/bc023h.pdf
Inactivation of a gene	Inactivación del gen	Gen-Inaktivierung	http://www.analesdepediatria.org/es-pdf-S1695403308720526
Inadequate suction	Succión inadecuada	unzureichendes Saugen	http://www.medigraphic.com/pdfs/abc/bc-

			2002/bc023h.pdf
Lack of speech	Capacidad de habla mínima	minimale Sprechkapazität	http://www.medigraphic.com/pdfs/abc/bc-2002/bc023h.pdf
Mandibular prognathism	Prognatismo	Prognathie	http://www.coem.org.es/sites/default/files/revista/cientifica/vol7-n1/61-64.pdf
Methylation testing	Análisis de metilación	Methylierungs-Analyse	http://www.analesdepediatria.org/es-pdf-S1695403308720526
Microcephaly	Microcefalia	Mikrozephalie	http://www.medigraphic.com/pdfs/abc/bc-2002/bc023h.pdf
Motor problema	Problema motor	motorische Störung	http://www.medigraphic.com/pdfs/abc/bc-2002/bc023h.pdf
Oral manifestation	Manifestación oral	mündliche Äußerung	http://www.coem.org.es/sites/default/files/revista/cientifica/vol7-n1/61-64.pdf
Paroxysmal laughter	Risa paroxística	paroxysmales Lachen	http://www.medigraphic.com/pdfs/abc/bc-2002/bc023h.pdf
Severe mental delay	Retraso mental severo	schwere mentale Retardierung	http://www.medigraphic.com/pdfs/abc/bc-2002/bc023h.pdf
Speech impairment	Déficit del lenguaje	Sprachdefizit	http://www.analesdepediatria.org/es-pdf-S1695403308720526
The 15q11-13	Región 15q11-13	chromosomale Region 15q11-13	http://www.medigraphic.com/pdfs/abc/bc-2002/bc023h.pdf
Tongue thrusting	Protrusión lingual	Protrusion der Zunge	http://www.coem.org.es/sites/default/files/revista/cientifica/vol7-n1/61-64.pdf
UB3A gene	Gen UB3A	UBE3A-Gen	http://www.analesdepediatria.org/es-pdf-S1695403308720526
Wide mouth	Boca amplia	breiter Mund	http://www.coem.org.es/site

			s/default/files/revista/cientifica/vol7-n1/61-64.pdf

2. Cerebral palsy (CP) in children, dystonic tetraparesis

ENGLISH	SPANISH	GERMAN	REFERENCES
Adulthood	Edad adulta	Erwachsenenalter	http://www.scielo.org.ve/scielo.php?script=sci_arttext&pid=S0004-06492013000100008
Associated disorder	Trastorno asociado	Begleiterkrankung	http://www.redalyc.org/pdf/3679/367937046008.pdf
Botulinum toxin	Toxina botulínica	Botulinumtoxin	http://www.elsevier.es/es-revista-neurologia-295-articulo-paralisis-cerebral-infantil-el-uso-S0213485315001516
Central Nervous System (CNS)	Sistema Nervioso Central (SNC)	zentrales Nervensystem (ZNS)	http://www.redalyc.org/pdf/3679/367937046008.pdf
Childhood disability	Discapacidad intantil	Behinderungen bei Kindern	http://www.medigraphic.com/pdfs/opediatria/op-2014/op141b.pdf
Clinical type	Tipo clínico	klinische Merkmale	http://www.medigraphic.com/pdfs/opediatria/op-2014/op141b.pdf
Consanguinity	Consanguinidad	Inzucht/Blutsverwandtschaft	http://www.redalyc.org/pdf/3679/367937046008.pdf
Defects of posture	Defectos de la postura	Haltungsschäden	http://www.redalyc.org/pdf/3679/367937046008.pdf
Deformity	Deformidad	Deformität	http://www.elsevier.es/es

			-revista-neurologia-295-articulo-paralisis-cerebral-infantil-el-uso-S0213485315001516
Disabled person	Minusválido	behinderte Person	http://www.revistapediatria.cl/volumenes/2014/vol11num2/pdf/PARALISIS_CEREBRAL.pdf
First neuron	Primera neurona	1. Motoneuron (Synonym für oberes Motoneuron)	http://www.redalyc.org/pdf/3679/367937046008.pdf
Gestational age	Edad de gestación	Gestationszeit	http://www.scielo.org.ve/scielo.php?script=sci_arttext&pid=S0004-06492013000100008
Hip dislocation	Luxación de cadera	Hüftluxation	http://www.elsevier.es/es-revista-neurologia-295-articulo-paralisis-cerebral-infantil-el-uso-S0213485315001516
Infant cerebral palsy (ICP)	Parálisis cerebral infantile (PCI)	infantile Zerebralparese (ICP)	http://www.redalyc.org/pdf/3679/367937046008.pdf
Integral treatment	Tratamiento integral	ganzheitliche Behandlung	http://www.scielo.org.ve/scielo.php?script=sci_arttext&pid=S0004-06492013000100008
Lower motor neuron	Neurona motora inferior	unteres Motoneuron	http://www.scielo.org.ve/scielo.php?script=sci_arttext&pid=S0004-06492013000100008
Maternal malnutrition	Desnutrición materna	mütterliche Mangelernährung	http://www.redalyc.org/pdf/3679/367937046008.pdf
Metabolic disease	Enfermedad metabólica	Stoffwechselerkrankung	http://www.redalyc.org/pdf/3679/367937046008.p

			df
Motor disorder	Trastorno motor	motorische Störung	http://www.medigraphic.com/pdfs/opediatria/op-2014/op141b.pdf
Neurodegenerative disease	Enfermedad neurodegenerativa	neurodegenerative Erkrankung	http://www.redalyc.org/pdf/3679/367937046008.pdf
Neurodevelopmental tracing	Seguimiento neuroevolutivo	Überwachung der fortschreitenden neuronalen Entwicklung	http://www.redalyc.org/pdf/3679/367937046008.pdf
Neurology	Neurología	Neurologie	http://www.elsevier.es/es-revista-neurologia-295-articulo-paralisis-cerebral-infantil-el-uso-S0213485315001516
Neuropathological study	Estudio enuropatológico[1]	neuropathologische Studie [1][auf spanisch muss es *neuropatológico* heiβen]	http://www.redalyc.org/pdf/3679/367937046008.pdf
Orthoses	Ortesis	Orthese	http://www.elsevier.es/es-revista-neurologia-295-articulo-paralisis-cerebral-infantil-el-uso-S0213485315001516
Orthotic aid	Ayuda ortésica	orthopädische Hilfe	http://www.elsevier.es/es-revista-neurologia-295-articulo-paralisis-cerebral-infantil-el-uso-S0213485315001516
Pathogenesis	Etiopatogenia	Ätiopathogenese	http://www.redalyc.org/pdf/3679/367937046008.pdf
Physical	Discapacidad	motorische	http://www.redalyc.org/pdf/3679/367937046008.p

disability	motriz	Behinderung	df
Prenatal control	Control prenatal	pränatales Screening	http://www.redalyc.org/pdf/3679/367937046008.pdf
Respiratory sepsis	Sepsis respiratoria	Atemwegsvergiftung	http://scielo.sld.cu/pdf/amc/v12n4/amc02408.pdf
Sitting position	Posición de pedestación	Sitzhaltung	http://www.elsevier.es/es-revista-neurologia-295-articulo-paralisis-cerebral-infantil-el-uso-S0213485315001516
Social integration	Integración social	soziale Integration	http://www.revistapediatria.cl/volumenes/2014/vol11num2/pdf/PARALISIS_CEREBRAL.pdf
Standing position	Posición de bipedestación	Haltung beim Stehen	http://www.elsevier.es/es-revista-neurologia-295-articulo-paralisis-cerebral-infantil-el-uso-S0213485315001516
Syndromic	Sindromático	syndromatisch	http://www.revistapediatria.cl/volumenes/2014/vol11num2/pdf/PARALISIS_CEREBRAL.pdf
Therapeutic intervention	Intervención terapéutica	therapeutischer Eingriff	http://www.medigraphic.com/pdfs/opediatria/op-2014/op141b.pdf
Upper motor neuron	Neurona motora superior	oberes Motoneuron	http://www.redalyc.org/pdf/3679/367937046008.pdf

3. Cornelia de Lange syndrome

Anxiety	Ansiedad	Angst(zustände)	https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf
Augmentative	Sistemas de	unterstützte	https://ecplusproject.uma.es/sites

and Alternative Communication Systems (AAC systems)	Comunicación Aumentativos y Alternativos (Sistema de CAA)	Kommunikation (UK)	/default/files/guide-eng.pdf
Chromosome cohesion	Cohesión cromosómica	chromosomale Kohäsion	http://www.aeped.es/sites/default/files/documentos/sindrome_de_cornelia_de_lange.pdf
Cognitive impairment[1]	Retraso psicomotor[2]	[1](Übersetzung des engl. Terminus): cognitive Beeinträchtigung [2](Übersetzung des span. Terminus): Verzögerung in der psychomotorischen Entwicklung	http://www.aeped.es/sites/default/files/documentos/sindrome_de_cornelia_de_lange.pdf
Cohesin Complex	Complejo de Cohesinas	Cohesin-Komplex	http://www.aeped.es/sites/default/files/documentos/sindrome_de_cornelia_de_lange.pdf
Facial dysmorphia[1]	Fenotipo facial[2]	[1](Übersetzung des engl. Terminus): faziale Dysmorphie [2](Übersetzung des span. Terminus): fazialer Phänotyp	http://www.aeped.es/sites/default/files/documentos/sindrome_de_cornelia_de_lange.pdf
Fenotype	Fenotipo	Phänotyp	http://www.aeped.es/sites/default/files/documentos/sindrome_de_cornelia_de_lange.pdf
Gastroesophageal reflux	Reflujo gastroesofágico	gastro-ösophagealer Reflux	http://www.aeped.es/sites/default/files/documentos/sindrome_de_cornelia_de_lange.pdf
Gene expression	Expresión génica	Genexpression	https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf
Growth	Retraso del	[1](Übersetzung des engl. Terminus):	http://www.aeped.es/sites/default/files/documentos/sindrome_de

impairment[1]	crecimiento[2]	Wachstumsbeeinträchtigung [2](Übersetzung des span. Terminus): Verzögerung des Wachstums	_cornelia_de_lange.pdf
Heterogeneous disorder[1]	Trastorno del desarrollo hereditario[2]	[1](Übersetzung des engl. Terminus): heterogene Störung [2](Übersetzung des span. Terminus): erbliche Entwicklungsstörung	http://www.aeped.es/sites/default/files/documentos/sindrome_de_cornelia_de_lange.pdf
Hirsutism	Hirsutismo	Hirsutismus	https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf
Limb malformation[1]	Anomalía en las extremidades superiores[2]	[1](Übersetzung des engl. Terminus): Fehlbildung der Gliedmaßen [2](Übersetzung des span. Terminus): Anomalie der oberen Extremitäten	http://www.aeped.es/sites/default/files/documentos/sindrome_de_cornelia_de_lange.pdf
Mental retardation	Retraso mental	mentale Retardierung	http://www.aeped.es/sites/default/files/documentos/sindrome_de_cornelia_de_lange.pdf
Mild fenotype	Fenotipo leve	schwacher Phänotyp	http://www.aeped.es/sites/default/files/documentos/sindrome_de_cornelia_de_lange.pdf
Moderate fenotype	Fenotipo moderado	moderater Phänotyp	http://www.aeped.es/sites/default/files/documentos/sindrome_de_cornelia_de_lange.pdf
Mutation	Mutación	Mutation	https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf
NIPBL gene	Gen NIPBL	NIPBL-Gen	http://www.aeped.es/sites/default/files/documentos/sindrome_de_cornelia_de_lange.pdf
Oligodactylia	Oligodactilia	Oligodaktylie	https://ecplusproject.uma.es/sites

			/default/files/guide-eng.pdf
Psychomotor retardation	Retraso psicomotor	verzögerte psychomotorische Entwicklung	https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf
Regulatory protein	Proteína reguladora	regulierendes Protein	http://www.aeped.es/sites/default/files/documentos/sindrome_de_cornelia_de_lange.pdf
Self-harm	Autoagresión	selbstverletzendes Verhalten/Selbstverletzung	https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf
Severe fenotype	Fenotipo grave	starker Phänotyp	http://www.aeped.es/sites/default/files/documentos/sindrome_de_cornelia_de_lange.pdf
SMC1A gene	Gen SMC1A	SMC1A-Gen	http://www.aeped.es/sites/default/files/documentos/sindrome_de_cornelia_de_lange.pdf
SMC3 gene	Gen SMC3	SMC3-Gen	http://www.aeped.es/sites/default/files/documentos/sindrome_de_cornelia_de_lange.pdf
Structural protein	Proteína estructural	Strukturprotein	http://www.aeped.es/sites/default/files/documentos/sindrome_de_cornelia_de_lange.pdf
Synophridia	Sinofridia	Synophrys	https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf

4. Deafness associated with other causes of intellectual disability

ENGLISH	SPANISH	GERMAN	REFERENCES
"Tleacle" protein[1]	Proteína «tleacle»[1]	Treacle-Protein [1] es muss auf englisch und spanisch *treacle* heiβen	http://www.medigraphic.com/pdfs/pediat/sp-2010/sp104f.pdf
Autosomal-dominant pattern	Patrón de herencia autosómico-dominante	autosomal dominantes Vererbungsschema	http://www.medigraphic.com/pdfs/pediat/sp-2010/sp104f.pdf
Charge Syndrome	Síndrome de Charge	Charge-Syndrom	https://ecplusproject.uma.es/sites/default/files/guide-esp.pdf y https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf
Choanal atresia	Atresia de coanas	Choanalatresie	http://www.scielo.org.ar/pdf/aap/v108n1/v108n1a16.pdf
Chromodomain	Cromodominio	Chromodomäne	http://www.scielo.org.ar/pdf/aap/v108n1/v108n1a16.pdf
Coloboma	Coloboma	Kolobom	http://www.scielo.org.ar/pdf/aap/v108n1/v108n1a16.pdf
Congenital heart defect	Cardiopatía	Kardiopathie	http://www.scielo.org.ar/pdf/aap/v108n1/v108n1a16.pdf
Cranofacial deformity	Deformidad craneofacial	kranofaziale Deformität	http://www.medigraphic.com/pdfs/pediat/sp-2010/sp104f.pdf
Ear anomalies	Malformaciones auriculares	Fehlbildungen des Ohrs	http://www.scielo.org.ar/pdf/aap/v108n1/v108n1a16.pdf
Genital abnormalities	Genitales anómalos	Genitalanomalien	http://www.scielo.org.ar/pdf/aap/v108n1/v108n1a16.pdf

Goldenhar Syndrome	Síndrome Goldenhar	Goldenhar-Syndrom	https://ecplusproject.uma.es/sites/default/files/guide-esp.pdf y https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf
Hearing loss	Sordera	Gehörlosigkeit	http://www.scielo.org.ar/pdf/aap/v108n1/v108n1a16.pdf
Helicase	Helicasa	Helikase	http://www.scielo.org.ar/pdf/aap/v108n1/v108n1a16.pdf
Hemifacial microsomia	Microsomia hemifacial	hemifaziale Mikrosomie	https://ecplusproject.uma.es/sites/default/files/guide-esp.pdf y https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf
Heterogeneous disorder	Desorden heterogéneo	heterogene Störung	http://www.scielo.org.co/pdf/ceso/v28n1/v28n1a7.pdf
Hypertrichosis	Hipertricosis	Hypertrichosis	http://www.medigraphic.com/pdfs/pediat/sp-2010/sp104f.pdf
Hypoplasia renal	Hipoplasia renal derecha	rechte renale Hypoplasie	http://www.medigraphic.com/pdfs/pediat/sp-2010/sp104f.pdf
Macrostomy	Macrostomia	Makrostomie	http://www.medigraphic.com/pdfs/revciemedhab/cmh-2015/cmh152m.pdf
Maxillofacial	Maxilofacial	maxillofazial	http://www.scielo.org.co/pdf/ceso/v28n1/v28n1a7.pdf
Microty	Microtia	Mikrotie	http://www.medigraphic.com/pdfs/revciemedhab/cmh-2015/cmh152m.pdf
Oculo Auriculo Vertebral	Espectro Óculo Aurículo Vertebra	okulo-aurikulo-vertebrales Spektrum	http://www.scielo.org.co/pdf/ceso/v28n1/v28n1

spectrum			a7.pdf
Pharyngeal arch	Arco faríngeo	Pharynxbogen	http://www.medigraphic.com/pdfs/pediat/sp-2010/sp104f.pdf
Severe micrognaty	Micrognatia severa	schwere Mikrognathie	http://www.medigraphic.com/pdfs/revciemedhab/cmh-2015/cmh152m.pdf
Treacher-Collins Syndrome	Síndrome de Treacher Collins	Treacher-Collins-Syndrom	https://ecplusproject.uma.es/sites/default/files/guide-esp.pdf y https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf
Zygoma	Cigoma	Zygoma/Jochbein	http://www.medigraphic.com/pdfs/revciemedhab/cmh-2015/cmh152m.pdf

5. Infantile epileptic encephalopathies: Dravet syndrome

ENGLISH	**SPANISH**	**GERMAN**	**REFERENCES**
Adverse effect	Reacción adversa	Nebenwirkung	http://www.medigraphic.com/pdfs/imss/im-2011/im111i.pdf
Anticonvulsants	Anticonvulsicante	Antikonvulsiva	http://www.medigraphic.com/pdfs/imss/im-2011/im111i.pdf
Brain abscence	Ausencia atípica	atypische Absencen	http://www.medigraphic.com/pdfs/imss/im-2011/im111i.pdf
Clinical evidence	Evidencia	(klinischer) Befund	http://www.medigraphic.com/pdfs/imss/im-2011/im111i.pdf
Clinical expression	Manifestación symptoms	klinische Ausprägung	http://www.medigraphic.com/pdfs/imss/im-

			2011/im111i.pdf
Contraindication	Contraindicación	Kontraindikation	http://www.medigraphic.com/pdfs/imss/im-2011/im111i.pdf
Cryptogenic	Criptogénico	kryptogen	http://www.medigraphic.com/pdfs/imss/im-2011/im111i.pdf
Diagnostic guide	Abordaje diagnóstico	diagnostischer Ansatz	http://www.medigraphic.com/pdfs/imss/im-2011/im111i.pdf
Entiepileptic drug	Antiepiléptico	Antikonvulsiva	http://www.medigraphic.com/pdfs/imss/im-2011/im111i.pdf
Epileptic syndrome	Síndrome epiléptico	epileptisches Syndrom	http://www.medigraphic.com/pdfs/imss/im-2011/im111i.pdf
Hereditary antecedent	Antecedente hederofamiliar	erbliche Vorgeschichte	http://www.medigraphic.com/pdfs/imss/im-2011/im111i.pdf
Identify a causal	Causa identificada	identifizierte Ursache	http://www.medigraphic.com/pdfs/imss/im-2011/im111i.pdf
Idiopathic	Idiopático	idiopathisch	http://www.medigraphic.com/pdfs/imss/im-2011/im111i.pdf
Infant group	Preescolar	Vorschulalter	http://www.medigraphic.com/pdfs/imss/im-2011/im111i.pdf
Juvenile myoclonic epilepsy	Epilepsia mioclónica juvenil	myoklonische Epilepsie bei Kindern, auch: juvenile myoklonische Epilepsie	http://www.medigraphic.com/pdfs/imss/im-2011/im111i.pdf
Neonatal group	Recién nacido	Neugeborener/-nes	http://www.medigraphic.com/pdfs/imss/im-

			2011/im111i.pdf
Neonatal symptomatic seizure	Crisis neonatal sintomática	neonataler symptomatischer Krampfanfall	http://www.medigraphic.com/pdfs/imss/im-2011/im111i.pdf
Paraclinic manifestation	Manifestación paraclínica	paraklinische Befunde	http://www.medigraphic.com/pdfs/imss/im-2011/im111i.pdf
Pharmalogical interaction	Interacción farmacológica	Arzneimittelwechselwirkung	http://www.medigraphic.com/pdfs/imss/im-2011/im111i.pdf
Pronostic	Pronóstico	Prognose	http://www.medigraphic.com/pdfs/imss/im-2011/im111i.pdf
Scholar group	Escolares	Grundschulalter/Schulkinder	http://www.medigraphic.com/pdfs/imss/im-2011/im111i.pdf
Seizure	Crisis	Krampfanfälle	http://www.medigraphic.com/pdfs/imss/im-2011/im111i.pdf
Symptomatic	Sintomático	symptomatisch	http://www.medigraphic.com/pdfs/imss/im-2011/im111i.pdf
The International League against Epilepsy (ILAE)	Liga Internacional Contra la Epilepsia	Internationale Liga gegen Epilepsie	http://www.medigraphic.com/pdfs/imss/im-2011/im111i.pdf
Therapeutic dose	Dosis terapéutica	Behandlungsdosis	http://www.medigraphic.com/pdfs/imss/im-2011/im111i.pdf
Therapeutic guide	Abordaje terapéutico	therapeutischer Ansatz	http://www.medigraphic.com/pdfs/imss/im-2011/im111i.pdf

6. Kleefstra syndrome

ENGLISH	SPANISH	GERMAN	REFERENCES
Aggressive outbursts	Arrebato de agresividad	aggressive Ausbrüche	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Apathy	Apatía	Apathie	https://www.ncbi.nlm.nih.gov/books/NBK47079/
Attention deficit problems	Déficit de atención	Aufmerksamkeitsdefizitstörungen	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Autosomal dominant transmission	Transmisión autosómica dominante	autosomal dominante Vererbung	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Brachy-microcephaly	Braqui-microcefalia	Brachy-Mikrozephalie	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Cardiac anomaly	Anomalía cardíaca	Herzfehler/Herzanomalie	https://www.ncbi.nlm.nih.gov/books/NBK47079/
Catatonia	Catatonia	Katatonie	https://www.ncbi.nlm.nih.gov/books/NBK47079/
Characteristic facial appearance	Reconocible apariencia facial	erkennbare Gesichtszüge	https://www.ncbi.nlm.nih.gov/books/NBK47079/
Chromosome	Cromosoma	Chromosom	https://www.ncbi.nlm.nih.gov/books/NBK47079/

Chromosome region	Región cromosómica	Chromosomenregion	https://www.ncbi.nlm.nih.gov/books/NBK47079/
Clinical description	Descripción clínica	klinische Beschreibung	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Connective tissue disorders	Trastorno del tejido conectivo	Bindegewebserkrankung	https://www.kleefstrasyndrome.org/what-is-kleefstra-syndrome/
Constipation	Estreñimiento	Obstipation	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Craniofacial abnormality	Dismorfia craneofacial	kraniofaziale Dysmorphie	https://www.ncbi.nlm.nih.gov/pubmed/28057753
Cupid's bow	Arco de Cupido	Amorbogen	https://www.ncbi.nlm.nih.gov/books/NBK47079/
Deletion	Deleción	Deletion	https://www.kleefstrasyndrome.org/what-is-kleefstra-syndrome/
Dental anomalies	Anomalía dental	Zahnanomalie	https://www.ncbi.nlm.nih.gov/books/NBK47079/
Developmental disorder	Trastorno del desarrollo	Entwicklungsstörung	https://www.kleefstrasyndrome.org/what-is-kleefstra-syndrome/
Emotional outbursts	Arrebato emocional	emotionale Ausbrüche	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&E

			xpert=261494
Enzyme	Encima	Enzym	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Épidemiology	Epidemiología	Epidemiologie	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Epilepsy	Epilepsia	Epilepsie	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Etiology	Etiología	Ätiologie/Ursachen	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Euchromatic histone--lysine N-methyltransferase 1 (*EHMT1*)	Gen histona-lisina N-metiltransferasa 1 eucromática *(EHMT1)*	Gen der euchromatischen Histon-Lysin-N-Methyltransferase 1 (*EHMT1*)	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Facial appearance	Rasgos faciales	Gesichtszüge	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Fluorescent in situ hybridization *(FISH)*	Hibridación fluorescente in situ *(FISH)*	fluoreszierende In-Situ Hybridisierung (FISH)	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Full evertid lower lip	Labio inferior evertido	evertierte Unterlippe	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494

Genetic disorder	Trastorno genético	genetische Störung	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Genital defects	Defectos genitales	Genitalfehlbildungen	https://www.ncbi.nlm.nih.gov/books/NBK47079/
Hearing problems	Discapacidad auditiva	Schwerhörigkeit	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Hearing problems	Problema de audición	Schwerhörigkeit	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Hypotonia	Hipotonía	Hypotonie	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Intellectual disability	Discapacidad intelectual	geistige Behinderung	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Intestinal monitoring	Monitorización intestinal	Darmkontrolle/intestinale Überwachung	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Intragenic insertion	Duplicación intragénica	intragenische Vervielfältigung	https://www.ncbi.nlm.nih.gov/books/NBK47079/
Kleefstra syndrome (KS)	Síndrome de Kleefstra (SK)	Kleefstra-Syndrome (KS)	https://www.kleefstrasyndrome.org/what-is-kleefstra-syndrome/

Low muscle tone	Bajo tono muscular	geringer Muskeltonus	https://www.kleefstrasyndrome.org/what-is-kleefstra-syndrome/
Microarray	Microarray	Mikroarray	https://www.ncbi.nlm.nih.gov/books/NBK47079/
Microdeletion	Microdeleción	Mikrodeletion	https://www.ncbi.nlm.nih.gov/books/NBK47079/
Midface hipoplasia	Hipoplasia del tercio medio facial	Mittelgesichtshypoplasie	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Molecular genetic testing	Test genético molecular	molekularer Gentest	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Motor delay	Retraso motor	eine Verzögerung in der motorischen Entwicklung	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Multiplex ligation-dependent probe amplification	Amplificación múltiple dependiente de sonda *(MLPA)*	[Die Abkürzung MLPA steht für:] Multiplex Ligation-dependent Probe Amplification	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Overweight	Sobrepeso	Übergewicht	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Physical and occupational therapy	Ergoterapia	Ergotherapie	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494

Physical therapy	Fisioterapia	Physiotherapie	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Primary dentition	Dentición primaria	Milchzähne	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Prognathism	Prognatismo	Prognathie	https://www.ncbi.nlm.nih.gov/books/NBK47079/
Protruding tongue	Lengua prominente	Protrusion der Zunge	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Recurrent infection	Infección recurrente	wiederkehrende Infektionen	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Renal abnormalities	Defectos renales	Nierenstörungen	https://www.ncbi.nlm.nih.gov/books/NBK47079/
Respiratory insufficiency	Complicación respiratoria	Atemschwierigkeiten	https://www.ncbi.nlm.nih.gov/books/NBK47079/
Seizures	Convulsiones	Konvulsion/Krampf	https://www.ncbi.nlm.nih.gov/books/NBK47079/
Self-mutilation	Autolesión	selbstverletzendes Verhalten/Selbstverletzungen	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Sensory integration therapy	Terapia de integración sensorial	sensorische Integrationstherapie	https://www.orpha.net/consor/cgi-bin/OC_Exp.p

			hp?lng=EN&Expert=261494
Sleep disturbances	Trastorno del sueño	Schlafstörungen	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Speech/language therapy	Logopedia	Sprachtherapie	https://www.ncbi.nlm.nih.gov/books/NBK47079/
Strabismus/Squint	Estrabismo	Schielen	https://www.kleefstrasyndrome.org/what-is-kleefstra-syndrome/
Structural brain abnormalities	Anomalías cerebrales estructurales	strukturelle Hirnanomalien	https://www.ncbi.nlm.nih.gov/books/NBK47079/
Synophyrs	Sinofridia	Synophrys	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494
Tracheomalacia	Traqueomalacia	Tracheomalazie	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=261494

7. Lennox Gastaut syndrome

ENGLISH	**SPANISH**	**GERMAN**	**REFERENCES**
Abnormal psychomotor development	Desarrollo psicomotor anormal	abnormale psychomotorische Entwicklung	https://books.google.es/books?id=7oEpBAAAQBAJ&pg=PA34&lpg=PA34&dq=%22abnormal+psychomotor+development%22+lennox&s

			ource=bl&ots=ibLu6bYUQP&sig=-7gNH7ls9GJty1yQX-iL8_h7oeY&hl=es&sa=X&ved=0ahUKEwj6o_eUt7fbAhWCOhQKHcmiAsoQ6AEILzAB#v=onepage&q=%22abnormal%20psychomotor%20development%22&f=false
Angelman syndrome	Síndrome de Angelman	Angelman-Syndrom	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Antiepileptic drugs	Fármacos antiepilépticos	Antikonvulsiva	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Atonic seizure	Crisis atónicas	atonische Krämpfe	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Atypical absences	Ausencias atípicas	atypische Absencen	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Axial tonic seizures	Crisis tónicas axiales	axial-tonische Krämpfe	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Benign partial epilepsy	Epilepsia parcial benigna	benigne partielle Epilepsie	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Cerebral pathology	Patología cerebral	Hirnschäden	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382

Ceroid lipofuscinoses	Lipofuscinosis ceroidea	Ceroid-Lipofuscinose	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Childhood epilepsy	Epilepsia infantil	Kindheitsepilepsie	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Conventional therapy	Terapia convencional	konventionelle Behandlungsmethoden	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Cortical dysplasia	Diplasia cortical	kortikale Dysplasie	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Cranial trauma	Trauma craneal	Hirnschädigung	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Cryptogenic epilepsy	Epilepsia criptogénica	kryptogenes Krampfleiden (generalisiertes Krampfleiden von unbekannter oder bekannter Ätiologie)	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Cryptogenic forms	Formas criptogénicas	kryptogene Formen	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Électroencephalogram (EEG)	Electroencefalograma (EEG)	Elektroenzephalogramm (EEG)	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Electroencephalographic	Electroencefalográfico	Elektroenzephalographie	https://www.ncbi.nlm.nih.gov/pubmed/1395056
Epileptic seizure	Crisis epilépticas	schwere Krampfanfälle	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
ESES síndrome	Síndrome ESES	ESES-Syndrom	https://www.orpha.net/consor/cgi-

			bin/OC_Exp.php?lng=EN&Expert=2382
Fast rhythmic bursts	Descargas rítmicas rápidas	schnelle rhythmische Entladungen	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Felbamate	Felbamato	Felbamat	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
General anaesthesia	Anestesia general	Vollnarkose	https://www.orpha.net/data/patho/Ans/en/Lennox-GastautSyndrome_DE_en_ANS_ORPHA2382.pdf
Intellectual déficit	Déficit intelectual	geistige Behinderung	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Lamotrigine	Lamotrigina	Lamotrigin	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Landau-Kleffener syndrome	Síndrome de Landau-Kleffener	Landau-Kleffner-Syndrom	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Lennox-Gastaut syndrome	Síndrome de Lennox-Gastaut (LGS)	Lennox-Gastaut-Syndrom (LSG)	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Levetiracetam	Levetiracetam	Levetiracetam	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Meningoencephalitis sequelae	Secuelas por meningoencefalitis	Folgen einer Meningoenzephalitis	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Mental health	Salud mental	geistige Gesundheit	https://www.scien

			cedirect.com/science/article/pii/S105913110900212X
Metabolic diseases	Enfermedades metabólicas	Stoffwechseldefekte	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Minor motor seizures	Crisis motoras menores	kurze epileptische Anfälle (sog. '*minor motor seizures*')	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Multifocal severe epilepsy	Epilepsia multifocal severa	multifokale schwere Epilepsie	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Myoclonic epilepsies	Epilepsias mioclónicas	myoklonische Epilepsien	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Neurological abnormalities	Trastornos neurológicos	neurologische Erkrankungen	https://ghr.nlm.nih.gov/condition/lennox-gastaut-syndrome
Perinatal asphyxia	Asfixia perinatal	perinatale Asphyxie	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Personality disturbances	Alteraciones de la personalidad	Persönlichkeitsveränderungen	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Polymorphism	Polimorfismo	Polymorphismus	https://www.ejpn-journal.com/article/S1090-3798(17)31087-5/fulltext?code=yejpn-site
Pre-existent brain damage	Daños cerebrales preexistentes	schon bestehender Hirnschaden	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Rett syndrome	Síndrome de Rett	Rett-Syndrom	https://www.orpha.net/consor/cgi-

			bin/OC_Exp.php?lng=EN&Expert=2382
Rufinamide	Rufinamida	Rufinamid	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Seizure disorder	Trastorno convulsivo	konvulsive Anfall-Störung	http://www.childneurologyfoundation.org/disorders/lgs-lennox-gastaut-syndrome/
Slow interictal spike wave	Punta-onda interictal lenta	langsame [interiktale] Spike-Waves/Slow-Spike-Waves	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Slow mental development	Desarrollo mental lento	verzögerte geistige Entwicklung	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Status epilepticus	Status epilepticus	*Status epilepticus*	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Sudden atonic or myoclonic falls	Caídas repentinas atónicas o mioclónicas	plötzliche atonische/astatische oder myoklonische Sturzanfälle	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Symptomatic generalized epilepsy	Epilepsia sintomática	generalisiertes Krampfleiden	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Symptomatic triad	Tríada sintomática	Symptomen-Trias	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382
Thyrotropin-releasing hormone (TRH)	Hormona liberadora de tirotropina (TRH)	Thyrotropin-freisetzendes Hormon (TRH)	https://www.ncbi.nlm.nih.gov/pubmed/11814729
Topiramate	Topiramato	Topiramat	https://www.orpha.net/consor/cgi-

			bin/OC_Exp.php?lng=EN&Expert=2382
Tuberous sclerosis	Esclerosis tuberosa	tuberöse Sklerose	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2382

8. MECP2: Rett syndrome

ENGLISH	SPANISH	GERMAN	REFERENCES
Apnea	Apnea	Apnoe	http://www.revistasbolivianas.org.bo/scielo.php?pid=S2304-37682014000700004&script=sci_arttext
Apraxia	Apraxia	Apraxie	http://www.revistasbolivianas.org.bo/scielo.php?pid=S2304-37682014000700004&script=sci_arttext
Ataxia	Ataxia	Ataxie	http://www.revistasbolivianas.org.bo/scielo.php?pid=S2304-37682014000700004&script=sci_arttext
Autism	Autismo	Autismus	http://www.revistasbolivianas.org.bo/scielo.php?pid=S2304-37682014000700004&script=sci_arttext

Behavioural change	Cambio conductual	Änderung des Verhaltens	http://www.scielo.org.ar/scielo.php?script=sci_arttext&pid=S0025-76802007000700002
Brain maturation	Maduración cerebral	Gehirnreifung	http://www.revistasbolivianas.org.bo/scielo.php?pid=S2304-37682014000700004&script=sci_arttext
CDKL5 gene	Gen CDKL5	CDKL5-Gen	http://www.scielo.org.ar/scielo.php?script=sci_arttext&pid=S0025-76802007000700002
Cerebral palsy	Parálisis cerebral	Zerebralparese	http://scielo.sld.cu/scielo.php?script=sci_arttext&pid=S1727-897X2012000100013
Child psychomotor impairment	Deterioro psicomotriz infantil	psychomotorische Beeinträchtigung des Kindes	http://www.revistasbolivianas.org.bo/scielo.php?pid=S2304-37682014000700004&script=sci_arttext
Head growth	Crecimiento craneal	Schädelwachstum/ Kopfwachstum	http://www.revistasbolivianas.org.bo/scielo.php?pid=S2304-37682014000700004&script=sci_arttext

Infantile autism	Autismo infantil	Autismus bei Kindern	http://scielo.sld.cu/scielo.php?script=sci_arttext&pid=S1727-897X2012000100013
Infantile dementia	Demencia infantil	infantile Demenz lat. *dementia infantilis*	http://www.revistasbolivianas.org.bo/scielo.php?pid=S2304-37682014000700004&script=sci_arttext
Irregular breathing	Alteración de la respiración	unregelmäßige Atmung	http://www.scielo.org.ar/scielo.php?script=sci_arttext&pid=S0025-76802007000700002
Pseudo stationary stage	Etapa pseudoestacionaria	Plateauphase	http://www.revistasbolivianas.org.bo/scielo.php?pid=S2304-37682014000700004&script=sci_arttext
Psychomotor development	Desarrollo psicomotor	psychomotorische Entwicklung	http://www.revistasbolivianas.org.bo/scielo.php?pid=S2304-37682014000700004&script=sci_arttext
Psychomotor skill	Destreza psicomotora	psychomotorische Fähigkeit	http://scielo.sld.cu/scielo.php?script=sci_arttext&pid=S1727-897X2012000100013

Regression stage of development	Etapa de regresión del desarrollo	Phase der Regression	http://www.revistasbolivianas.org.bo/scielo.php?pid=S2304-37682014000700004&script=sci_arttext
Scoliosis	Escoliosis	Skoliose	http://www.revistasbolivianas.org.bo/scielo.php?pid=S2304-37682014000700004&script=sci_arttext
Seizure	Crisis epiléptica	Krampfanfälle	http://scielo.sld.cu/scielo.php?script=sci_arttext&pid=S1727-897X2012000100013
Social disorder	Alteración social	soziale Störung	http://scielo.sld.cu/scielo.php?script=sci_arttext&pid=S1727-897X2012000100013
Spasticity	Espasticidad	Spastik	http://www.revistasbolivianas.org.bo/scielo.php?pid=S2304-37682014000700004&script=sci_arttext
Stage of late motor deterioration	Etapa de deterioro motor tardío	Phase der motorischen Verschlechterung	http://www.revistasbolivianas.org.bo/scielo.php?pid=S2304-37682014000700004&script=sci_arttext

Stage of stagnation	Etapa de estancamiento	Phase der Stagnation	http://www.revistasbolivianas.org.bo/scielo.php?pid=S2304-37682014000700004&script=sci_arttext
Stereotype	Estereotipia	Stereotyp	http://www.scielo.org.ar/scielo.php?script=sci_arttext&pid=S0025-76802007000700002
X chromosome (MECP2)	Cromosoma X (MECP2)	X Chromosom (MECP2)	http://www.revistasbolivianas.org.bo/scielo.php?pid=S2304-37682014000700004&script=sci_arttext

9. Microdelation and microduplication syndromes with serious language affectation

ENGLISH	**SPANISH**	**GERMAN**	**REFERENCES**
Arch palate	Paladar ojival	spitzer Gaumen	https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf
Autosomal dominant	Autosómica dominante	autosomal-dominant	https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf
Chromosome 1	Cromosoma 1	Chromosom 1	https://ecplusproject.uma.es/sites/default/files/gui

			de-eng.pdf
Comparative genomic hybridization	Hibridación genómica comparativa	vergleichende Genom-Hybridisierung (CGH)	https://ecpluspro ject.uma.es/sites /default/files/gui de-eng.pdf
Deletion	Deleción	Deletion	https://ecpluspro ject.uma.es/sites /default/files/gui de-eng.pdf
Dental malocclusion/De ntal oclusal disharmony	Maloclusión dental	dentale Malokklusion	https://ecpluspro ject.uma.es/sites /default/files/gui de-eng.pdf
Dolichocephaly	Dolicocefalia	Dolichozephalie	https://ecpluspro ject.uma.es/sites /default/files/gui de-eng.pdf
Facial dimorphism	Dimorfismo facial	faziale Dysmorphie	https://ecpluspro ject.uma.es/sites /default/files/gui de-eng.pdf
Hypotony	Hipotonía	Hypotonie	https://ecpluspro ject.uma.es/sites /default/files/gui de-eng.pdf
Inbred	Consanguíneo	blutsverwandt	https://ecpluspro ject.uma.es/sites /default/files/gui de-eng.pdf
Microcephaly	Microcefalia	Mikrozephalie	https://ecpluspro ject.uma.es/sites /default/files/gui de-eng.pdf
Moderate deficit	Déficit moderado	leichte Behinderung	https://ecpluspro ject.uma.es/sites /default/files/gui

			de-eng.pdf
Novo gene mutation	Mutación de novo	Neumutation	https://ecpluspro ject.uma.es/sites /default/files/gui de-eng.pdf
Phenotype	Fenotipo	Phänotyp	http://www.anal esdepediatria.or g/es-sndrome-microdelecin-22q11-manifestaciones-cardiorrespirator ias-articulo-S169540331200 0951
Retrognathia	Retrognatia	Retrognathie	https://ecpluspro ject.uma.es/sites /default/files/gui de-eng.pdf
Severe speech delay	Retardo grave (en el lenguajeo)	schwere verzögerte Sprachentwicklung	https://ecpluspro ject.uma.es/sites /default/files/gui de-eng.pdf

10. Mowat-Wilson syndrome

ENGLISH	**SPANISH**	**GERMAN**	**REFERENCES**
Agenesis of corpus callosum	Agenesia del cuerpo calloso	Agenesie des Corpus callosum	https://www.orp ha.net/consor/cg i-bin/OC_Exp.ph p?lng=EN&Exp ert=2152
Arterial duct	Canal arterial	arterierer Kanal	https://www.ncb i.nlm.nih.gov/m edgen/4415
Atypical phenotype	Fenotipo atípico	atypische Symptomatik	https://www.orp ha.net/consor/cg

			i-bin/OC_Exp.php?lng=EN&Expert=2152
Autosomal dominant	Autosómico dominante	autosomal-dominant	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Cardiac anomalies	Anomalías cardiacas	Herzfehler/Herzanomalie	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Central depression	Depresión central	[große, aufwärts gerichtete Ohrläppchen mit] zentraler Einsenkung	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Chromosome 23	Cromosoma 23	Chromosom 23	http://www.ivami.com/en/genetic-testing-human-gene-mutations-diseases-neoplasias-and-pharmacogenetics/4240-genetic-testing-mowat-wilson-syndrome-mowat-wilson-syndrome-gen-i-zeb2-i
Clinical features	Rasgos clínicos	klinische Merkmale	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Congenital malformations	Defectos congénitos	angeborene Fehlbildungen	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Delayed	Retraso en el	verzögerte psychomotorische	https://www.orp

psychomotor development	desarrollo psicomotor	Entwicklung	ha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Digestive tract	Tracto digestivo	Verdauungstrakt	http://www.ivami.com/en/genetic-testing-human-gene-mutations-diseases-neoplasias-and-pharmacogenetics/4240-genetic-testing-mowat-wilson-syndrome-mowat-wilson-syndrome-gen-i-zeb2-i
Distinct facial phenotype	Fenotipo facial distintivo	distinktiver fazialer Phänotyp	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Dysmorphic facial features	Dismorfia facial	faziale Dysmorphie	http://www.mowatwilsonsyndrome.org.uk/profile8.htm
Early clinical investigation	Estudio clínico temprano	klinische Früherkennungsstudie/ mit rehabilitativen Maßnahmen (Physiotherapie, psychomotorische und Sprachtherapie) sollte daher so früh wie möglich begonnen werden	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Embryonic development	Desarrollo embrionario	embryonale Entwicklung	http://www.ivami.com/en/genetic-testing-human-gene-mutations-diseases-neoplasias-and-pharmacogenetics/4240-genetic-testing-mowat-wilson-syndrome-mowat-wilson-syndrome-gen-i-

			zeb2-i
Endocrine glands	Hormonas endocrinas	endokrine Hormone	http://www.ivami.com/en/genetic-testing-human-gene-mutations-diseases-neoplasias-and-pharmacogenetics/4240-genetic-testing-mowat-wilson-syndrome-mowat-wilson-syndrome-gen-i-zeb2-i
Épilepsy	Epilepsia	Epilepsie	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Genitourinary anomalies	Anomalías genitourinarias	Urogenital-Anomalien	hhttps://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Haploinsufficiency	Haploinsuficiencia	Haploinsuffizienz	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Heterozygous mutations	Deleciones heterocigotas	heterozygote Mutationen	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Hirschsprung disease	Enfermedad de Hirschsprung	Hirschsprung-Krankheit (HSCR)	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Hirschsprung disease	Síndrome de Hirschsprung	Hirschsprung-Krankheit (HSCR)	https://www.orpha.net/consor/cgi-

			bin/OC_Exp.php?lng=EN&Expert=2152
Hypertelorism	Hipertelorismo	Hypertelorismus	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Hypospadias	Hipospadias	Hypospadie	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Intellectual disability	Discapacidad intelectual	geistige Behinderung	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Intestinal disorder	Alteración intestinal	Darmerkrankung/intestinale Erkrankungen	http://www.ivami.com/en/genetic-testing-human-gene-mutations-diseases-neoplasias-and-pharmacogenetics/4240-genetic-testing-mowat-wilson-syndrome-mowat-wilson-syndrome-gen-i-zeb2-i
Intestinal obstruction	Obstrucción intestinal	Darmverschluss	http://www.ivami.com/en/genetic-testing-human-gene-mutations-diseases-neoplasias-and-pharmacogenetics/4240-genetic-testing-mowat-wilson-syndrome-mowat-wilson-syndrome-gen-i-

			zeb2-i
Mecanisme physiopathologique	Mecanismo patológico	[physio]pathologischer Mechanismus	https://www.ncbi.nlm.nih.gov/pubmed/12189494
Microcephaly	Microcefalia	Mikrozephalie	http://www.ivami.com/en/genetic-testing-human-gene-mutations-diseases-neoplasias-and-pharmacogenetics/4240-genetic-testing-mowat-wilson-syndrome-mowat-wilson-syndrome-gen-i-zeb2-i
Mowat-Wilson syndrome (MWS)	Síndrome de Mowat-Wilson (MWS)	Mowat-Wilson-Syndrom (MWS)	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Multiple congenital anomaly syndrome	Síndrome de anomalías congénitas múltiples	'Multiple-congenital-anomaly'-Syndrom	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Mutational analysis	Análisis mutacional	Mutationsanalyse	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Mutations	Mutaciones	Mutationen	http://www.ivami.com/en/genetic-testing-human-gene-mutations-diseases-neoplasias-and-pharmacogenetics/4240-genetic-testing-mowat-wilson-syndrome-

			mowat-wilson-syndrome-gen-i-zeb2-i
Nasal bridge	Puente nasal	Nasenrücken	http://www.ivami.com/en/genetic-testing-human-gene-mutations-diseases-neoplasias-and-pharmacogenetics/4240-genetic-testing-mowat-wilson-syndrome-mowat-wilson-syndrome-gen-i-zeb2-i
Neonatologist	Neonatólogo	Neonatologe	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Neural crest	Cresta neural	Neuralleiste	http://www.ivami.com/en/genetic-testing-human-gene-mutations-diseases-neoplasias-and-pharmacogenetics/4240-genetic-testing-mowat-wilson-syndrome-mowat-wilson-syndrome-gen-i-zeb2-i
Ocular anomalies	Anomalías oculares	Augenanomalien	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Ophtalmologic abnormalities	Trastornos oftalmológicos	ophthalmologische Erkrankungen	https://www.ncbi.nlm.nih.gov/pubmed/22486326
Prominent columella	Columela prominente	prominente Columella	https://www.orpha.net/consor/cg

			i-bin/OC_Exp.php?lng=EN&Expert=2152
Rare disease	Enfermedad rara	seltene Krankheit	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Rectal biopsy	Biopsia rectal	Rektumbiopsie	http://www.mowatwilsonsyndrome.org.uk/mowat-wilson-syndrome.htm
Saddle nose	Nariz en silla de montar	Sattelnase	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Seizures	Convulsiones	Krampfleiden	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Severe constipation	Extreñimiento grave	schwere Obstipation	http://www.ivami.com/en/genetic-testing-human-gene-mutations-diseases-neoplasias-and-pharmacogenetics/4240-genetic-testing-mowat-wilson-syndrome-mowat-wilson-syndrome-gen-i-zeb2-i
Skeletal muscle	Músculos esqueléticos	Skelettmuskulatur	http://www.ivami.com/en/genetic-testing-human-gene-mutations-diseases-neoplasias-and-pharmacogeneti

			cs/4240-genetic-testing-mowat-wilson-syndrome-mowat-wilson-syndrome-gen-i-zeb2-i
Smooth muscle	Músculo liso	glatte Muskulatur	http://www.ivami.com/en/genetic-testing-human-gene-mutations-diseases-neoplasias-and-pharmacogenetics/4240-genetic-testing-mowat-wilson-syndrome-mowat-wilson-syndrome-gen-i-zeb2-i
Studies of genotype-phenotype	Estudios de análisis genotipo-fenotipo	Genotyp-Phänotyp-Analyse	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Underdiagnosed	Infradiagnosticado	Die Prävalenz [des MWS] ist noch nicht bekannt	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Variable congenital malformations	Malformaciones congénitas variables	verschiedene angeborene Fehlbildungen	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
ZEB2 protein	Proteína ZEB2	ZEB2-Protein	http://www.ivami.com/en/genetic-testing-human-gene-mutations-diseases-neoplasias-and-pharmacogenetics/4240-genetic-testing-mowat-

			wilson-syndrome-mowat-wilson-syndrome-gen-i-zeb2-i
Zinc finger E-box-binding homeobox 2 gene (ZEB2)	Gen homeobox 2 ligador de E-box que contiene un dedo de zinc (ZEB2)	Zinkfinger E-box-bindende Homeobox 2 Gen (ZEB2)	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152

11. Phelan-McDermid syndrome

ENGLISH	**SPANISH**	**GERMAN**	**REFERENCES**
Absent speech	Ausencia de lenguaje oral	fehlende Sprachentwicklung	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=48652
Absent to severely delayed speech	Retraso o ausencia de adquisición del lenguaje	fehlender oder stark verzögerter Spracherwerb	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=48652
Autism	Autismo	Autismus	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Autism spectrum disorder	Trastorno del espectro autista	Autismus-Spektrum-Störung (ASD)	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=48652
Bulbous nose	Nariz bulbosa	knollige Nase	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Cerebral palsy	Parálisis cerebral	Zerebralparese	https://ojrd.biomedcentral.com/ar

			ticles/10.1186/1750-1172-3-14
Chromosome analysis	Análisis cromosómico	Chromosomenanalyse	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=48652
Chromosome deletions	Deleciones en los cromosomas	Chromosomen-Deletion	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Chromosome microdeletion	Microdeleción cromosómica	Chromosomen-Mikrodeletion	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Comparative genomic hybridization (CGH)	Hibridación genómica comparativa (HGC)	vergleichende Genom-Hybridisierung (CGH)	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Cryptic rearrangements	Reordenamientos cromosómicos crípticos	kryptische Chromosomen-Umlagerungen	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Differential diagnosis	Diagnóstico diferencial	Differentialdiagnose	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Dolicocephaly	Dolicocefalia	Dolichozephalie	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Dysplastic nails	Uñas displásicas	dysplastische Zehennägel	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Fluorescence *in situ* diagnostic (FISH)	Hibridación fluorescente *in situ* (FISH)	Fluoreszenz-*in situ*-Hybridisierung (FISH)	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Gastrointestinal problems	Problemas gastrointestinales	gastrointestinale Probleme/Magen-Darm-Beschwerden	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=48652
Genetic material	Material genético	Erbgut	https://ojrd.biomedcentral.com/articles/10.1186/1

			750-1172-3-14
Hypotonia	Hipotonía	Hypotonie	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Infant stimulation programs	Programas de intervención tempranas	Programme zur Frühförderung/ frühe Interventionsprogramme	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Inherited rearrangements	Reordenamientos cromosómicos hereditarios	vererbte Chromosomen-Umlagerungen	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Karyotype	Cariotipo	Karyotyp	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Learning disabilities	Trastorno de aprendizaje	Lernstörungen	https://ldaamerica.org/types-of-learning-disabilities/
Microdeletion	Microdeleción	Mikrodeletion	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Minor dysmorphic features	Rasgos dismórficos menores	geringfügige Dysmorphien	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Monosomy 22q13	Monosomía 22q13	Monosomie 22q13	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Neonatal hypotonia	Hipotonía neonatal	Neonatale Hypotonie	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Neurological problems	Problemas neurológicos	neurologische Erkrankungen	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=48652
Normal to accelerated growth	Crecimiento normal o acelerado	normales oder akzeleriertes Wachstum	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Parental mosaicism	Mosaicismo parental	elterliches Mosaik	https://ojrd.biomedcentral.com/ar

			ticles/10.1186/1750-1172-3-14
Phelan-McDermid syndrome	Síndrome de Phelan-McDermid	Phelan-McDermid-Syndrom	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=48652
Pointed chin	Barbilla puntiaguda	spitzes Kinn	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Prenatal diagnosis	Diagnóstico prenatal	prenatale Diagnostik	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Primary physician	Médico generalista	Allgemeinarzt/Hausarzt	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Renal problems	Problemas renales	nephrologische Erkrankungen	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=48652
Ring chromosome	Cromosoma en anillo	Ringchromosom	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
SHANK3 gene	Gen SHANK3	*SHANK3*-Gen	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Sport programs	Terapias deportivas	Sportprogramme	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Structural abnormalities	Anomalías estructurales	strukturelle Anomalien	https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-3-14
Synapses	Sinapsis	Synapsen	https://www.ncbi.nlm.nih.gov/pubmed/25894671
Global developmental delay	Retraso global del desarrollo	allgemeine Entwicklungsverzögerung	https://www.orpha.net/consor/cgi-bin/OC_Exp.ph

			p?lng=EN&Expert=48652
Translocation	Translocación	Translokation	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=48652
Trichorhinophalangeal syndrome	Síndrome tricorrinofalángico	trichorhinophalangeales Syndrom	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=48652
Underdiagnosed síndrome	Síndrome infradiagnosticado	selten diagnistiziertes Syndrom	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=48652
Velocardiofacial síndrome	Síndrome velocardiofacial	velokardiofaziales Syndrom	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=48652

12. Pitt-Hopkins syndrome

ENGLISH	**SPANISH**	**GERMAN**	**REFERENCES**
Amniocentesis	Amniocentesis	Amniozentese	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Angelman síndrome	Síndrome de Angelman	Angelman-Syndrom	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Anomalies	Anomalías	Anomalien	https://www.ncbi.nlm.nih.gov/books/NBK100240

			/
Apnea	Apnea	Apnoe	https://www.ncbi.nlm.nih.gov/books/NBK100240/
Autosomal dominant transmission	Transmisión autosómica dominante	autosomal-dominante Vererbung	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Behavioural problems	Problemas de conducta	Verhaltensprobleme	http://pitthopkins.org.uk/?page_id=92
b-HLH transcription factor	Factor de transcripción b-HLH ubicuo	ubiquitärer b-HLH-Transkriptionsfaktor	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Brain scans	TACs cerebrales	kraniale CT [Computertomografie]	http://pitthopkins.org.uk/?page_id=92
Breathing abnormalities	Patrón de respiración anormal	abnorme und unregelmäßige Atmung	http://pitthopkins.org.uk/?page_id=92
Colour change	Cambios de colorido	Farbveränderungen	http://pitthopkins.org.uk/?page_id=92
Constipation	Estreñimiento	Obstipation	http://pitthopkins.org.uk/?page_id=92
Coordination of their limbs	Coordinación de las articulaciones	Koordination der Gliedmaßen	http://pitthopkins.org.uk/?page_id=92
Corpus callosum	Cuerpo calloso	*Corpus callosum*	http://pitthopkins.org.uk/?page_id=92
Cryptorchidism	Criptorquidia	Kryptorchismus	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Cyanosis	Cianosis	Zyanose	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896

Dedifferentiation of the temporal lobes	Desdiferenciación de los lóbulos temporales	Dedifferenzierung der Temporallappen	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Deep-set eyes	Ojos hundidos	tiefliegende Augen	http://pitthopkins.org.uk/?page_id=92
Delayed motor development	Retardo en el desarrollo motor	verzögerte motorische Entwicklung	http://pitthopkins.org.uk/?page_id=92
Differential diagnosis	Diagnóstico diferencial	Differentialdiagnose	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Ears with wide hélices	Orejas con hélices grandes	breite Ohrhelix	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Epileptic attacks	Crisis epiléptica	epileptische Anfälle	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Facial dysmorphism	Dismorfia facial	faziale Dysmorphien	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Facial features	Rasgos faciales	Gesichtszüge	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Gastro-esophageal reflux	Reflujo gastro-esofágico	gastro-ösophagealer Reflux	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Heterozygous *de novo* mutations	Mutaciones de novo	heterozygote Neumutationen	https://www.orpha.net/consor/cg

	heterocigotas		i-bin/OC_Exp.php?lng=EN&Expert=2896
Hyperventilation	Hiperventilación	Hyperventilation	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Hypopigmented skin macules	Máculas cutáneas hipopigmentadas	hypopigmentierte Makula/auch: hypopigmierte Hautflecken	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Hypoplasia	Hipoplasia	Hypoplasie	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Hypotonia	Hipotonía	Hypotonie	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Inherited disorder	Afecciones hereditarias	Erbkrankheit	https://www.ncbi.nlm.nih.gov/books/NBK100240/
Intellectual déficit	Déficit intelectual	intellektuelles Defizit	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Intermittent periods	Ataques intermitentes	intermittierende Attacken	http://pitthopkins.org.uk/?page_id=92
Language therapist	Especialista del lenguaje	Sprachtherapeut	http://pitthopkins.org.uk/?page_id=92
Learning disability	Trastornos de aprendizaje	Lernstörungen	http://pitthopkins.org.uk/?page_id=92
Macrostomia	Macrostomía	Makrostomie	https://www.orpha.net/consor/cgi-

			bin/OC_Exp.php?lng=EN&Expert=2896
Malformations	Malformaciones	Fehlbildungen	https://www.ncbi.nlm.nih.gov/books/NBK100240/
Microcephaly	Microcefalia	Mikrozephalie	https://www.ncbi.nlm.nih.gov/books/NBK100240/
Mowat-Wilson síndrome	Síndrome de Mowat-Wilson	Mowat-Wilson-Syndrom	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Muscle tone	Tono muscular	Muskeltonus	http://pitthopkins.org.uk/?page_id=92
Myopia	Miopía	Myopie	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Parental somatic mosaicism	Mosaicismo somático parental	elterliches somatisches Mosaik	https://www.ncbi.nlm.nih.gov/books/NBK100240/
Pitt-Hopkins syndrome (PHS)	Síndrome de Pitt-Hopkins (PHS)	Pitt-Hopkins-Syndrom (PHS)	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Postnatal underdevelopment	Retraso en el crecimiento postnatal	postnatale Entwicklungsverzögerung	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Psychomotor disorders	Problemas psicomotores	psychomotorische Störungen	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Rare disease	Enfermedad rara	seltene Krankheiten	https://www.orpha.net/consor/cg

			i-bin/OC_Exp.php?lng=EN&Expert=2896
Reduced hippocampus	Hipocampo reducido	reduzierter Hippocampus	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Rett syndrome	Síndrome de Rett	Rett-Syndrom	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Spaced teeth	Dientes espaciados	mit großen Zahnabständen	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Specialist paediatric physiotherapist	Fisioterapeuta pediátrico especializado	Physiotherapeut/in mit dem Schwerpunkt Pädiatrie	http://pitthopkins.org.uk/?page_id=92
Specific disease	Enfermedad específica	spezifische Krankheit	https://www.ncbi.nlm.nih.gov/books/NBK100240/
Speech and language therapist	Logopeda	Logopäde/Logopädin	http://pitthopkins.org.uk/?page_id=92
Splayed nostrils	Fosas nasales separadas	ausgeweitete Nasenlöcher	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Symptoms	Síntomas	Symptome	http://pitthopkins.org.uk/?page_id=92
TCF4 gene (18q21)	Gen TCF4 (18q21)	*TCF4*-Gen (18q21)	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Unstable walking	Andar rígido	unstabiles Laufen	https://www.orpha.net/consor/cgi-

			bin/OC_Exp.php?lng=EN&Expert=2896
Ventricular dilation	Dilatación ventricular	erweiterter Ventrikel	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Voluntary prehension	Prensión voluntaria	willkürliches Greifen	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896
Wide palate	Paladar amplio	breiter und flacher Gaumen	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2896

13. Severe autism

ENGLISH	**SPANISH**	**GERMAN**	**REFERENCES**
Adjuvant	Adyuvante	unterstützend	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Anxiety	Ansiedad	Angst(zustände)	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Asymmetrical tonic neck reflex	Reflejo Tónico Asimétrico del Cuello	asymmetrischer Hals-Reflex	https://www.omicsonline.org/open-access/neurosensorimotor-reflex-integration-for-autism-a-new-

			therapymodality-paradigm-.php?aid=81336
Attention deficit hyperactivity disorder	Trastorno de déficit de atención	Aufmerksamkeitsdefizitstörung (ADHS)	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Autism	Autismo	Autismus	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Autism spectrum disorder (ASD)	Trastornos del espectro autista (TEA)	Autismus-Spektrum-Störungen (ASD)	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Behavioural treatment	Terapia conductual	Verhaltenstherapie	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Chronic noncommunicable conditions	Enfermedades no trasmisibles crónicas	chronische nichtübertragbare Krankheiten	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Co-occurring conditions	Afecciones comórbidas	Begleiterkrankungen	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Depression	Depresión	Depression	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Difficulties in communication	Dificultades de comunicación	Kommunikationsprobleme	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-

			disorders
Discrimination	Discriminación	Diskriminierung	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Emotional burden	Carga emocional	emotionale Belastung	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Epidemiology	Epidemiología	Epidemiologie	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Epilepsy	Epilepsia	Epilepsis	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
General developmental disorder	Trastorno generalizado del desarrollo	generelle Entwicklungsstörung	https://dc.uwm.edu/cgi/viewcontent.cgi?referer=https://www.google.es/&httpsredir=1&article=1777&context=etd
General health-care needs	Necesidades sanitarias generales	allgemeiner Bedarf an sanitären Diensten	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Health-care needs	Necesidades asistenciales	Pflegebedarf	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Human rights violations	Violación de los derechos humanos	Menschenrechtsverletzungen	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders

Humanitarian emergency	Emergencia humanitaria	humanitärer Notstand	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Hyperactivity disorder	Hiperactividad	Hyperaktivität	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Hypersensitivity	Hipersensibilidad	Überempfindlichkeit	http://www.autism.org.uk/sensory
Intellectual disability	Discapacidad intelectual	geistige Behinderung	http://www.intellectualdisability.info/diagnosis/articles/autism
Level of intelectual functioning	Nivel intelectual	intellektuelles Niveau	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Management	Conducta clínica	Behandlung/Therapie	http://www.who.int/fr/news-room/fact-sheets/detail/autism-spectrum-disorders
Measles, mumps and rubella vaccine	Vacuna contra el sarampión, parotiditis y rubéola	Impfung gegen Masern, Mumps und Röteln	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Mental retardation	Retraso mental	mentale Retardierung	https://www.asatonline.org/research-treatment/clinical-corner/mental-retardation-and-autism/
Methodological flaws	Errores metodológicos	methodische Mängel	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-

			disorders
Moro reflex	Reflejo de Moro	Moro-Reflex	https://www.omicsonline.org/open-access/neurosensorimotor-reflex-integration-for-autism-a-new-therapymodality-paradigm-.php?aid=81336
Neubiological disease	Enfermedad neurobiológica	neurobiologische Erkrankung	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Neurological disease	Trastorno neurológico	neurologische Erkrankung	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Physical inactivity	Inactividad física	körperliche Inaktivität	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Poor dietary preferences	Malas preferencias dietéticas	schlechte Essensgewohnheiten	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Profound impairment	Deterioro profundo	tiefgreifende Beeinträchtigungen	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Psychosocial interventions	Intervenciones psicosociales	psychosoziale Interventionen	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Severe autism	Autismo severo	schwerer Autismus	https://www.autismparentingmag

			azine.com/low-functioning-autism/
Severe behavioural disorder	Trastorno severo del comportamiento	schwere Verhaltensstörung	https://www.researchgate.net/publication/247890488_Autism_and_anorexia_nervosa_Related_conditions
Severe disabilities	Discapacidades graves	schwere Behinderungen	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Severe sensory abnormalities	Trastorno severo de la sensorialidad	schwere Sinnesstörungen	https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4918912/
Skills training programmes	Programas de capacitación	Trainingsprogramme	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Social behaviour	Comportamiento social	Sozialverhalten	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Stigma	Estigmatización	Stigmatisierung	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Superior intellectual functioning	Aptitudes cognitivas altas	überdurchschnittliche kognitive Fähigkeiten	https://www.tandfonline.com/doi/abs/10.1080/02783199909553992
Thiomersal	Tiomersal	Thimerosal	http://www.who.int/news-room/fact-sheets/detail/autism-spectrum-disorders
Treatment of	Tratamiento de	Behandlung akuter und chronischer	http://www.who.

acute and chronic illness	enfermedades agudas y crónicas	Erkrankungen	int/news-room/fact-sheets/detail/autism-spectrum-disorders

14. Usher syndrome

ENGLISH	**SPANISH**	**GERMAN**	**REFERENCES**
Acoustic trauma	Traumatismo acústico	akustisches Trauma	https://www.hear-it.org/glossary/a?name=
Acoustic tumour	Tumor acústico	akustisches Neuroma	https://www.hear-it.org/glossary/a?name=
Autosomal recessive	Autosómico recesivo	autosomal-rezessiv	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=886
Cataracts	Catarata	Katarakt/Grauer Star	https://nei.nih.gov/health/ushers/ushers
Chromosome	Cromosoma	Chromosom	https://www.nature.com/articles/ejhg201115.pdf
Cochlea	Cóclea	Cochlea	https://www.hear-it.org/glossary/c?name=
Cones	Conos	Konus/Conus	https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4482614/
Congenital deafness	Sordera congénita	angeborene Schwerhörigkeit	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=886
Crystalline lens	Cristalino	Augenlinse	https://www.eye

			glossary.net/#C
Daytime vision	Visión diurna	Sehvermögen	https://www.hear-it.org/usher-s-syndrome
Deafness	Sordera	Taubheit	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=886
Electroretinogram (ERG)	Electro-retinograma	Elektroretinogramm	https://www.nature.com/articles/ejhg201115.pdf
Endolymph	Endolinfa	Endolymphe	https://www.ncbi.nlm.nih.gov/pubmedhealth/PMHT0028178/
Genetic diagnostics	Diagnóstico genético	Gendiagnostik	https://www.nature.com/articles/ejhg201115.pdf
Genetic disorder	Enfermedad genética	genetische Erkrankung	https://www.usher-syndrome.org/what-is-usher-syndrome/frequent-questions.html
Gradual vision loss	Pérdida visual progresiva	fortschreitender Sehverlust	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=886
Hearing	Audición	Hörvermögen	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=886
Hearing aid	Audífonos	Hörgeräte	https://www.hear-it.org/glossary/h?name=
Hearing impaired	Discapacitado auditivo	Hörgeschädigter	http://www.nchearingloss.org/hi.htm
Inner ear	Oído interno	Innenohr	https://www.hear-it.org/glossary/i?name=

Language acquisition	Adquisición del lenguaje	Spracherwerb	https://www.usher-syndrome.org/our-story/blog/sign-or-not-3-avt.html
Myopia	Miopía	Myopie	https://nei.nih.gov/eyedata/myopia
Night blindness	Ceguera nocturna	Nachtblindheit	https://nei.nih.gov/health/ushers/ushers
Night vision	Visión nocturna	nächtliches Sehvermögen	https://www.nidcd.nih.gov/health/usher-syndrome
Ophthalmologist	Oftalmólogo	Ophthalmologe	https://www.eyeglossary.net/#C
Ophthalmoscopy	Oftalmoscopia	Ophthalmoskopie	https://medlineplus.gov/ency/article/003881.htm
Optic nerve	Nervio óptico	*Nervus opticus* (Sehnerv)	https://www.eyeglossary.net/#C
Otoacoustic emissions	Otoemisiones acústicas (OEA)	otoakustische Emissionen (OAE)	https://www.sciencedirect.com/topics/medicine-and-dentistry/usher-syndrome
Partial deafness	Pérdida parcial de la audición	teilweiser Verlust der Hörfähigkeit	https://www.sciencedirect.com/science/article/pii/B9780123838346001476
Peripheral visión	Visión periférica	peripheres Sehen	https://www.nidcd.nih.gov/health/usher-syndrome
Photoreceptor	Fotorreceptores	Fotorezeptoren/Photorezeptoren (auch Sehzellen)	https://www.sciencedirect.com/topics/medicine-and-dentistry/usher-syndrome
Poor balance	Escaso equilibrio	schlechter Gleichgewichtssinn	https://www.usher-syndrome.org/what-is-usher-

			syndrome/frequent-questions.html
Presbyacusis	Presbiacusia	Presbyakusis (Altersschwerhörigkeit)	https://www.hear-it.org/glossary/p?name=
Rare disease	Enfermedad poco común	seltene Krankheit	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=886
Recessive gene	Gen recesivo	rezessives Gen	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=886
Retina	Retina	Retina (Netzhaut)	https://www.sciencedirect.com/topics/medicine-and-dentistry/usher-syndrome
Retinitis pigmentosa	Retinitis pigmentosa	*Retinitis pigmentosa*	https://www.sciencedirect.com/science/article/pii/B9780128053980000074
Rods	Bastones	Stäbchen	https://www.ncbi.nlm.nih.gov/pubmed/28495838
Sensorineural deafness	Sordera sensorineural	Schallempfindungs-Schwerhörigkeit	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=886
Sign language	Lenguaje de signos	Gebärdensprache	https://www.usher-syndrome.org/what-is-usher-syndrome/frequent-questions.html
Specialist	Médico especialista	Facharzt	https://www.orpha.net/consor/cgi-bin/OC_Exp.ph

			p?lng=EN&Expert=886
Speech spectrum	Espectro del habla	Sprachspektrum	https://www.hear-it.org/glossary/s?name=
Tinnitus	Acúfenos	Tinnitus	https://www.hear-it.org/glossary/t?name=tinnitus
Total deafness	Sordera total	vollständige Gehörlosigkeit	https://www.sciencedirect.com/science/article/pii/B9780123838346001476
Usher syndrome	Síndrome de Usher	Usher-Syndrom (US)	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=886
Vestibular areflexia	Ausencia de función vestibular	vestibuläre Areflexie	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=886
Vestibular disorders	Trastornos vestibulares	vestibuläre Störungen	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=886
Vestibulocochlear nerve	Nervio auditivo	Hörnerv (*Nervus vestibulocochlearis*)	https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3872449/
Visual field	Campo visual	Gesichtsfeld	https://nei.nih.gov/health/ushers/ushers

15. West's syndrome

ENGLISH	SPANISH	GERMAN	REFERENCES
Adrenocorticotropic hormone (ACTH)	Hormona corticotropina (ACTH)	adrenocorticotropes Hormon (ACTH)	http://www.revistachilenadeepilepsia.cl/wp-content/uploads/2015/04/201403_actualizacion_sindrome_west.pdf
Catastrophic epilepsy	Epilepsia catastrófica	verheerende/katastrophale Epilepsie	http://www.revistachilenadeepilepsia.cl/wp-content/uploads/2015/04/201403_actualizacion_sindrome_west.pdf
Clinic suspicion	Sospecha clínica	klinischer Verdacht	https://www.siis.net/documentos/ficha/217518.pdf
Cryptogenic	Criptogénico	kryptogen	http://www.redalyc.org/pdf/3250/325038650007.pdf
Differential diagnosis	Diagnóstico diferencial	Differentialdiagnose	http://www.redalyc.org/pdf/3250/325038650007.pdf
EEG (electroencephalogram)	Electroencefalograma	Elektroenzephalogramm (EEG)	https://www.siis.net/documentos/ficha/217518.pdf
Electroclinical triad	Tríada electroclínica	elelektroklinische Symptomen-Trias	http://www.revistachilenadeepile

			psia.cl/wp-content/uploads/2015/04/201403_actualizacion_sindrome_west.pdf
Epileptic spasm	Espasmo epiléptico	epileptische Spasmen	http://www.redalyc.org/pdf/3250/325038650007.pdf
Etiology	Etiología	Ätiologie/Ursachen	http://www.redalyc.org/pdf/3250/325038650007.pdf
Exitus	*Exitus*	*Exitus*	https://www.siis.net/documentos/ficha/217518.pdf
Hyperbilirubinemia	Hiperbilirrubinemia	Hyperbilirubinämie	http://www.medigraphic.com/pdfs/imi/imi-2012/imi121b.pdf
Hypsarrhythmia	Hipsarritmia	Hypsarrhythmie	http://www.redalyc.org/pdf/3250/325038650007.pdf
Idiopathic	Idiopático	idiopathisch	http://www.redalyc.org/pdf/3250/325038650007.pdf
Infantile spasm	Espasmo infantil	infantile Spasmen	http://www.redalyc.org/pdf/3250/325038650007.pdf
Mental impairment	Deterioro mental	mentale Beeinträchtigungen	http://www.medigraphic.com/pdfs/imi/imi-

			2012/imi121b.pdf
Myoclonic jerk	Crisis miclónica	myoklonischer Krampfanfall	http://www.medigraphic.com/pdfs/imi/imi-2012/imi121b.pdf
Neonatal pathology	Patología neonatal	neonatale Pathologie	http://www.medigraphic.com/pdfs/imi/imi-2012/imi121b.pdf
Neurodevelopment	Desarrollo neurológico	neurologische Entwicklung	http://www.redalyc.org/pdf/3250/325038650007.pdf
Paroxystic episode	Episodio paroxístico	paroxysmal	https://www.siis.net/documentos/ficha/217518.pdf
Pathophysiology	Fisiopatología	Pathophysiologie	http://www.redalyc.org/pdf/3250/325038650007.pdf
Pediatric age-dependent epileptic encephalopathy	Encefalopatía epiléptica pediátrica	epileptische Enzephalopatie bei Kindern	http://www.redalyc.org/pdf/3250/325038650007.pdf
Perinatal	Perinatal	perinatal	http://www.redalyc.org/pdf/3250/325038650007.pdf
Perinatal asphyxia	Asfixia perinatal	perinatale Asphyxie	http://www.medigraphic.com/pdfs/imi/imi-2012/imi121b.pdf

Postnatal	Posnatal	postnatal	http://www.redalyc.org/pdf/3250/325038650007.pdf
Prenatal	Prenatal	vorgeburtlich/pränatal	http://www.redalyc.org/pdf/3250/325038650007.pdf
Refractory	Refractorio*	refraktär *refractario (esp.)?!	http://www.revistachilenadeepilepsia.cl/wp-content/uploads/2015/04/201403_actualizacion_sindrome_west.pdf
Salaam tic	Tic de Salaam	Blitz-Nick-Salaam-Epilepsie/BNS-Epilepsie	http://www.redalyc.org/pdf/3250/325038650007.pdf
Seizure	Convulsión	Konvulsion/Krampf	http://www.revistachilenadeepilepsia.cl/wp-content/uploads/2015/04/201403_actualizacion_sindrome_west.pdf
Sepsis	Sepsis	Sepsis	http://www.medigraphic.com/pdfs/imi/imi-2012/imi121b.pdf
Spasms in clusters	Espasmos en salva	in Serien auftretende Spasmen	https://www.siis.net/documentos/ficha/217518.pdf
Symptomatic	Sintomático	symptomatisch	http://www.redalyc.org/pdf/3250

			/325038650007.pdf
Tuberous sclerosis	Esclerosis tuberosa	tuberöse Sklerose	http://www.medigraphic.com/pdfs/imi/imi-2012/imi121b.pdf
Vigabatrin	Vigabatrina	Vigabatrin	http://www.revistachilenadeepilepsia.cl/wp-content/uploads/2015/04/201403_actualizacion_sindrome_west.pdf

7. Multimodality in the subtitling of interactive tales: The case of Babel with EC+

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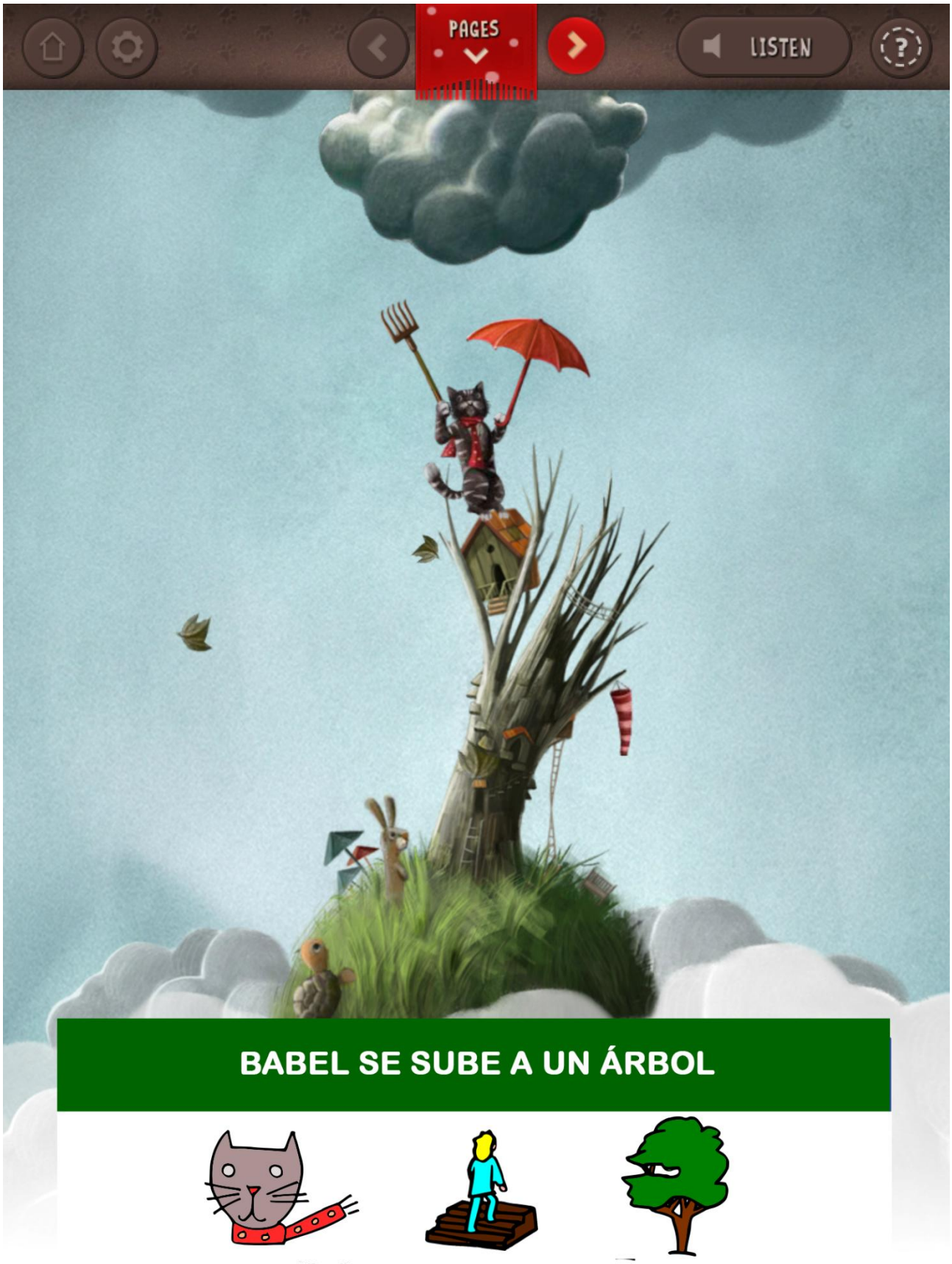
University of Málaga

The interactive tale subtitled and adapted with EC+ pictograms, *Babel the cat who would be king* by the Belgian company EPIC, is a pioneering multimodal EC+ resource, supported by an empirical study aimed at encouraging the development of communication skills within the framework of the European Erasmus EC+ Enhancing communication project. EC+ follows the principles of Augmentative and Alternative Communication (AAC) for teachers, caregivers and professionals who have to deal with people with Complex Communication Needs (CCN) due to physical (reduced mobility), sensory (visual or auditory) or cognitive (intellectual) disabilities. The interactive tale Babel was proposed to a total of 10 children, of which 5 are ASDs, 2 Down syndrome, 1 Idis 15 and 2 TEL. The results have been variable according to the cognitive abilities of the children. The diferent sequences of the tale are followed in the cases in which the cognitive ability is more preserved, although, in some cases, the actions are followed but without causality. The children who had the most difculties were children with ASDs. Some remarkable words were not well understood, such as, “grumpy, bored, says that a king needs a castle, who, adventures”. The most shocking images were when the lightning struck: everyone understood this event even though they do not understood the concept of the weather phenomenon. Similarly, another moment when almost everyone react was when the cat fell from the tree, children dropped to the ground. When asked, who wants to build a castle, most children said yes. The pictograms were understood according to levels, in some cases, there are some that have not understood when the complexity of the story increases, however, the basic emotions were understood. According to levels of cognition, children can relate what is most relevant in the story, although without following the order of the actions. We must take into account the fact that all of them have the expressive component of language and difculties since verbal memory was afected. Asking for the favorite character, there's equality between the cat and the bird. All the children found it easier to access the reading or simply visual comprehension of the story thanks to EC+pictograms.


PAGES
LISTEN
BABEL ES UN GATO VIEJO Y GRUÑÓN


PAGES
LISTEN
SU CASA ES VIEJA Y BABEL SE ABURRE


PAGES
LISTEN
BABEL SE SUBE A UN ÁRBOL


PAGES
LISTEN
BABEL NO PUEDE TOCAR LAS NUBES Y SE ENFADA


PAGES
LISTEN
UN RAYO LE ALCANZA. BABEL SE CAE DEL ÁRBOL


PAGES
LISTEN
BABEL VE UN PÁJARO DENTRO DE UNA JAULA


PAGES
LISTEN
BABEL PIENSA QUE ES UNA CORONA DE REY

8. Iconographic application in subtitles as a tool to foster comprehension in deaf and hard of hearing children.

Esther Sedano Ruiz

According to the main objective of the project, we aim to improve interaction with people with special needs by using ICT tools, and in this case we focused on the deaf and hard of hearing children and the understanding of the audiovisual material using icons and different resources in subtitles.

This activity took place in Colegio La Purísima in Malaga, where we collected information from children between 9 and 12 years old. In the first place, we projected a short film subtitled and adapted to them, in order to do so we adapted and reduced the subtitles for its better understanding and we also used icons instead of some words to attract the attention and facilitate the understanding of the main story and sounds. After the projection, they filled a questionnaire on their habits regarding television and subtitles and also about the comprehension of the story. Most of them watched TV or films at home but not always using subtitles and most of them had implants or hearing aids, nevertheless they all communicated always by sign language.

As a conclusion, children in this activity presented some difficulties to understand specific information, however we can say that the use of icons replacing words helped them to understand better the story as well as some specific information. Therefore, increasing the number of icons and reducing even more written subtitles would be something to consider in further studies, since children also presented problems concerning literary skills and making the audiovisual material even more visual would foster a better comprehension of subtitles.

Click on this link to watch the video: https://ecplusproject.uma.es/sites/default/files/TiJean.mp4

9. Tratamiento jurídico-social de la discapacidad: breves referencias a su evolución histórica y principales retos actuales

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1. Breves referencias a su evolución histórica

En España, una nueva etapa de protección jurídica de las personas con discapacidad, comienza a partir de la entrada en vigor en 1978 de la vigente Constitución. En este sentido, Lorenzo García[4], sostiene que: "La moderna etapa de protección jurídica de las personas con discapacidad arranca tras el novedoso artículo 49 de la CE que, siguiendo el reciente precedente de la Constitución portuguesa, incluye el catálogo de principios rectores de la política social y económica de la Carta Magna a la protección de un colectivo, como el de las personas con discapacidad, que había estado legislativamente casi olvidado en la evolución de nuestro ordenamiento jurídico. El artículo 49 inaugura una nueva etapa de enorme potencialidad interpretativa y transformadora de la realidad del derecho español (…)". Precisamente, del mandato constitucional de este artículo 49 de la CE[5], vería la luz la Ley 13/1982, de 7 de abril, de integración social de las personas con discapacidad (LISMI). Esta Ley, supuso un importante avance en materia jurídica sobre la protección de los derechos de las personas con discapacidad[6], integrando *"el modelo médico-rehabilitador de atención a las personas con discapacidad"*, y llevando a cabo "(…) un imponente desarrollo reglamentario en materias de gran alcance tales como: las prestaciones económicas; la educación especial; la integración laboral y la eliminación de barreras, por citar algunos de sus principales avances en esta materia[7]".

Años posteriores, se aprobaría la Ley 51/2003 de igualdad de oportunidades no discriminación y accesibilidad universal, que incorporaba como novedad, el cambio significativo del *"modelo médico-rehabilitador[8]"*, por el *"modelo social[9]"*; y la Ley 49/2007, de 26 de diciembre, de infracciones y sanciones de la LIONDAU[10].

[4] LORENZO GARCÍA, R. (2014), "El nuevo Texto Refundido de la Ley General de Derechos de la Personas con Discapacidad", en AA.VV., Protección social: Seguridad Social y Discapacidad. Estudios en homenaje a Adolfo Jiménez, Ed. Cinca, págs. 293–294.

[5] Véase en relación a lo dispuesto en el artículo 53 de la CE.

[6] Adviértase que en aquella época el término usado era "minusválido".

[7] Ib. Pág. 294.

[8] Es el modelo médico que se asienta sobre la rehabilitación de las personas con discapacidad. Véase en PALACIOS RIZZO, A. (2008), "El modelo social de discapacidad: orígenes, caracterización y plasmación en

Por su parte, y en relación al desarrollo histórico-jurídico de la protección de las personas con discapacidad en el ámbito de la UE, será en el Tratado de Ámsterdam, que entraría en vigor en 1997, cuando aparece por primera vez la necesidad de que la UE aborde el tema de la discriminación por razón de discapacidad. En su art.13, se autorizaba a la UE a adoptar medidas para luchar contra la discriminación de las personas con discapacidad, siendo ésta, de hecho, la primera referencia específica de los Tratados a la *discapacidad*. Este artículo proporcionaba por primera vez una base para abordar situaciones de discriminación múltiple, mediante el establecimiento de una política y un enfoque legal común para los distintos motivos de discriminación.

En virtud precisamente del fundamento jurídico que proporcionaba el art.13 del Tratado de Ámsterdam, se adoptó la aprobación de la Directiva 2000/78, sobre la igualdad de trato en el empleo y la ocupación, mediante la cual se prohibía la discriminación por motivos de sexo, de origen racial o étnico, religión o convicciones, *discapacidad*, edad u orientación sexual. En 2001, se modifica el art.13 por el Tratado de Niza con el objeto de permitir la adopción de medidas de incentivos por mayoría cualificada del Consejo[11].

Con la entrada en vigor del Tratado de Lisboa[12] en 2009, se establece el carácter vinculante de la Carta de Derechos Fundamentales, que contiene un amplio catálogo de derechos, destacando la transversalidad[13] de la no discriminación por razón de discapacidad[14]. También conviene destacar la incorporación de la Convención al acervo comunitario y al derecho positivo de los Estados miembros, lo que supone un importante avance en materia de discapacidad a nivel europeo, sobre todo a través de futuros desarrollos jurisprudenciales[15].

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la Convención Internacional sobre los Derechos de las Personas con Discapacidad", Madrid, Cinca, pág. 66.

[9] El modelo social pone el énfasis en la rehabilitación de una sociedad, que ha de ser concebida y diseñada para hacer frente a las necesidades de todas las personas, gestionando las diferencias e integrando la diversidad. Para profundizar en este aspecto, véase PÉREZ BUENO, L. (2010), "Discapacidad, derecho y políticas de inclusión", Madrid, Cinca, págs., 83-84. También, sobre este asunto ver en PALACIOS RIZZO, A. (2008), "El modelo social de discapacidad……op.cit. , págs. 106-107.

[10] Esta ley, desplegó sus efectos múltiples sobre nuevas iniciativas legislativas tales como la Ley de autonomía personal y protección de la dependencia, entre otras. Ver sobre este asunto en, LORENZO GARCÍA, R. (2014), "El nuevo Texto Refundido de la Ley...op. cit. págs. 294 y ss.

[11] Las medidas legislativas seguían requiriendo aprobación por unanimidad de todos los Estados miembro.

[12] Desarrollado con mayor amplitud en DANS ÁLVAREZ DE SOTOMAYOR, L., "Libre circulación de personas tras el Tratado de Lisboa", Madrid, *Revista del Ministerio e Inmigración. Derecho social Internacional y Comunitario,* núm. 92, 2011, págs. 257–282.

[13] Cit. Supra nota 9.

[14] Quedando encuadrado tras la aprobación en el art. 19 TFUE.

[15] CABRA DE LUNA, M., "La protección de la discapacidad en el ámbito internacional: la Unión Europea", en AA.VV., Protección social: Seguridad Social y Discapacidad. Estudios en homenaje a Adolfo Jiménez, Ed. Cinca, 2014, págs. 288–389.

La última iniciativa política y legislativa orientada a la lucha contra la discriminación y la igualdad de trato de las personas con discapacidad, que adoptó la Comisión Europea en 2010, es la Estrategia Europea sobre Discapacidad 2010-2020[16]. Tiene como una de las principales prioridades la incorporación de las personas con discapacidad al mercado laboral. Igualmente se centra en la eliminación de barreras e identifica medidas a escala de la UE complementarias a las acciones a nivel nacional. También determina los mecanismos necesarios para aplicar la Convención. Asimismo se focaliza en otros aspectos fundamentales para la plena integración y la igualdad de trato de las personas con discapacidad.

Sin embargo, el gran hito en materia de protección de los derechos de las personas con discapacidad, se va a producir con la aprobación en 2006 de la Convención de las Naciones Unidas sobre los derechos de las personas con discapacidad (CDPD), ratificada en 2010 por la UE. Es el primer instrumento amplio de derechos humanos del siglo XXI, así como el primer tratado internacional de derechos humanos que se abre a la firma de organizaciones regionales de integración, como la propia Unión Europea[17]. Además de recoger los derechos de las personas con discapacidad, también contempla las obligaciones de los Estados de promover, proteger y asegurar tales derechos[18]. A partir de su entrada en vigor en enero de 2011, tanto los Estados miembros como la propia UE han comenzado a adaptar sus ordenamientos jurídicos a la nueva realidad[19].

La ratificación de esta Convención por España el 3 de diciembre de 2007 y su posterior entrada en vigor el 3 de mayo de 2008, llevaba aparejada, la adaptación y reforma de la normativa española sobre los derechos de las personas con discapacidad y ha tenido como

[16] Sobre los objetivos, en AA.VV., *"Des droits pour tous: contribution des organismos de lutte contre les discriminations à l'évaluation de la Stratégie 2010-2020 de L'Union Européenne en faveur des personnes handicapées. Un avis equinet*", Bruselas, *Equinet European network of equality bodies,* diciembre, 2014, también en BALLESTER PASTOR, M., "La lucha contra la discriminación en la Unión Europea", Madrid, *Revista del Ministerio e Inmigración. Derecho social Internacional y Comunitario,* núm. 92, 2011, págs. 207–255.

[17] *Ib*. 389. También puede consultarse sobre este asunto el mismo autor, en: "El empleo de las personas con discapacidad en el ordenamiento jurídico comunitario y nacional", en CASTILLO DÍAZ, M. (pr.), I Congreso Nacional sobre Empleo de las Personas con Discapacidad: Hacia la plena inclusión laboral, ISBN 978-84-7993-307-4, UNIA, 2016, pág. 9.

[18] Concretamente, la Convención establece como principios generales el respeto de la dignidad inherente a la persona, la autonomía individual, la independencia de cada ser humano, la no discriminación, la participación e inclusión plena y efectivas en la sociedad, la igualdad de oportunidades, el respeto por la diferencia y la aceptación de las personas con discapacidad como manifestación de la diversidad y la condición humana. *Vid.* CABRA DE LUNA, M., "La protección de la discapacidad…, op. c., pág. 390.

[19] Sin embargo, al día de hoy queda mucho por hacer, es necesario que la Comisión Europea desarrolle unas directrices que permiten el *mainstreaming* de la CDPD en toda la legislación europea y que revise todo el acervo comunitario actualmente vigente. No le corresponde únicamente a la UE, realizar su trabajo, también es imprescindible que los Estados miembros realicen sus tareas.

consecuencia inmediata la elaboración del Real Decreto Legislativo 1/2013, de 29 de noviembre, por el que se aprueba el Texto Refundido de la Ley General de derechos de las personas con discapacidad y de su inclusión social –en adelante LGD-. Este Real Decreto, deroga expresamente -por refundirlas en este texto- las siguientes leyes: Ley 13/1982, de 7 de abril, de integración social de las personas con discapacidad (LISMI); Ley 51/2003, de 2 de diciembre, de igualdad de oportunidades, no discriminación y accesibilidad universal de las personas con discapacidad (LIONDAU) y Ley 49/2007, de 26 de diciembre, por la que se establece el régimen de infracciones y sanciones en materia de igualdad de oportunidades, no discriminación y accesibilidad universal de las personas con discapacidad. En referencia a la Igualdad de oportunidades y no discriminación, se describen las medidas contra la discriminación y las medidas de acción positiva. Se incluye, también por primera vez, las definiciones de todos los tipos de discriminación, directa e indirecta, ya contempladas, a las que se añaden la discriminación por asociación y acoso[20], con el fin de completar el marco jurídico de prohibición de la discriminación en cualquiera de sus manifestaciones[21]. Si bien la nueva ley no añade cambios sustanciales, sin duda simplifica y clarifica la dispersión existente hasta ahora en las leyes derogadas, y elimina las posibles contradicciones conceptuales que pudieran producirse entre textos tan dispares y alejados en el tiempo.

Por otra parte, conviene señalar (aunque de manera muy breve), que de forma paralela al desarrollo histórico-legislativo, se ha podido apreciar en el *ámbito jurisprudencial* (tanto a nivel nacional, como en el de la UE)[22], una evolución importante en materia de protección de los derechos de estas personas y a la no discriminación por razón de discapacidad.

A nivel global, en los últimos tiempos el tratamiento de la discapacidad, se ha convertido en un tema recurrente y de especial atención. Un examen sobre el contenido del último *Informe Mundial sobre la Discapacidad*[23], nos desvela un panorama desalentador

[20] En este sentido el artículo 27.1, a) de la Convención Internacional sobre los derechos de las personas con discapacidad. También en derecho comunitario, la Directiva 2000/43/CE sobre igualdad de trato de las personas independiente de su origen racial o étnico, en su artículo 2.3; Directiva 2000/78/CE sobre igualdad en el empleo, en su artículo 2.3 y la Directiva 2006/54/CE sobre igualdad de trato entre hombres y mujeres en asuntos de empleo y ocupación, en su artículo 2.1(c). En jurisprudencia comunitaria STJUE, de 17 de julio de 2008, asunto C-303/06, caso Coleman.

[21] En relación a la no discriminación en las relaciones laborales, el artículo 17 del ET.

[22] Para profundizar sobre este tema, puede consultarse en: RUIZ SANTAMARÍA, J.L. (2016), "La adaptación …op. cit. págs. 882-888. También en: MÁRQUEZ PRIETO, A. y RUIZ SANTAMARÍA, J.L. (2016), "Incapacidad total para un gruista con visión monocular al hilo de la STS de 23 de diciembre de 2014", Revista de derecho de la seguridad social, núm. extra 1 (dedicado a: Doctrina judicial en materia de Seguridad Social: balance y análisis selectivo de sentencias del año 2015), págs. 135-141.

[23] Este informe de fecha 9 de junio de 2011, emitido conjuntamente por la OMS y el Grupo del Banco Mundial y que lleva por título: "Informe mundial sobre la discapacidad: rompiendo barreras [*New World report on disability: Breakind down barriers*]", reúne la mejor información disponible sobre la

reflejándolo en los siguientes términos: "Más de mil millones de personas viven en todo el mundo con alguna forma de discapacidad; de ellas, casi doscientos millones experimentan dificultades considerables en su funcionamiento. En los años futuros, la discapacidad será un motivo de preocupación aún mayor, pues su prevalencia está aumentando. Ello se debe a que la población está envejeciendo y el riesgo de discapacidad es superior entre los adultos mayores, y también al aumento mundial de enfermedades crónicas tales como la diabetes, las enfermedades cardiovasculares, el cáncer y los trastornos de la salud mental". El citado informe sigue diciendo que: "(...) En todo el mundo, las personas con discapacidad tienen peores resultados académicos, ello es consecuencia de los obstáculos que entorpecen el acceso de las personas con discapacidad a servicios indispensables, en particular la salud, la educación, el empleo, el transporte, o la información". Estos perores resultados académicos, son el reflejo de los impedimentos y barreras[24] que se presentan para este colectivo, y que supone un lastre importante.

2. Principales retos actuales

A pesar de los avances legislativos y jurisprudenciales en esta materia, este colectivo tiene aún muchas dificultades[25], no solo para integrarse en la sociedad, sino que una vez integradas, puedan disfrutar de los mismos derechos que el resto de las personas[26]. En este sentido, un análisis del *"Informe Olivenza 2017, sobre la situación general de la discapacidad en España"*, nos ayudará a conocer de forma efectiva y real, cuáles son los retos actuales en materia de discapacidad.

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discapacidad con el fin de mejorar la vida de las personas que la padecen. También, ofrece a los gobiernos y a la sociedad civil un análisis exhaustivo de la importancia de la discapacidad y de las respuestas proporcionadas, basado en las mejores pruebas disponibles, y recomendar la adopción de medidas de alcance nacional e internacional. La visión que nos impulsa, es la de un mundo inclusivo en el que todos podamos vivir una vida de salud, comodidad y dignidad.

[24] Véase también en este sentido, LORENZO GARCÍA, R., "El futuro de los discapacitados en el mundo: el empleo como factor determinante para la inclusión", Madrid, *Revista del Ministerio e Inmigración,* núm. 50, 2004, págs. 73-90. El autor de este artículo ha dirigido por mandato del Club de Roma a nivel mundial, el informe titulado: "El futuro de las personas con discapacidad en el mundo. Desarrollo humano y discapacidad", en el que se realiza un profundo análisis del conjunto de problemas que afectan a la discapacidad a nivel mundial, desde la perspectiva de la integración y el reconocimiento pleno de los derechos inherentes a la dignidad de la persona, y desde la óptica del respeto a la diversidad. A través de la experiencia y aportaciones del referido informe, el autor aborda en este trabajo específico para la Revista del Ministerio de Trabajo y Asuntos Sociales la cuestión concreta de la inserción laboral de las personas con discapacidad como factor determinante para una plena integración social.

[25] Ib. pág. 880.

[26] En este sentido, *vid.* Informe Olivenza sobre la situación de la discapacidad en España (2014) del Observatorio estatal de la discapacidad, http://www.odismet.es/es/biblioteca/informe-olivenza-2014-sobre-la-discapacidad-en-espana/103/. (Consulta: 06/ 05/2016). Igualmente, Estrategia española sobre la discapacidad (2012 – 2020), http://sid.usal.es/libros/discapacidad/26112/8-4-1/estrategia-espanola-sobre-discapacidad-2012-2020.aspx. (Consulta: 01/07/2016) y su Plan de acción (2014 – 2016), http://www.lamoncloa.gob.es/consejodeministros/referencias/documents/2014/refc20140912e_7.pdf. (Consulta: 01/ 07/2016).

2.1 Situación sobre la inclusión social de las personas con discapacidad en España

Respecto a la inclusión social, la desventaja que presentan las personas con discapacidad, se cuantifica en un 65,4%.

La inclusión social como expresión máxima del ejercicio de los derechos de la ciudadanía con discapacidad que aborda cinco dimensiones fundamentales: participación política, educación, vivienda, salud y apoyos por discapacidad (este último específico y por tanto no comparable con la población general. En este sentido, y según los últimos indicadores, las personas con discapacidad alcanzan una distancia cercana al 55,1% con el resto de la población.

En cuanto a la *participación política*, el art. 29 CDPD, garantiza los derechos políticos entre los que se incluye el derecho al voto. Sin embargo, en clara contradicción con nuestra legislación interna, el art. 3 de la Ley 5/1985 de Régimen Electoral General, establece que los declarados incapaces en virtud de sentencia judicial firme quedarán privados del derecho del sufragio, siempre que la sentencia declare expresamente la incapacidad para su ejercicio. En este sentido, las personas con discapacidad que estuvieron privadas de su derecho al voto en las elecciones generales de 20 de diciembre de 2015 y 26 de junio de 2016, asciende a un total de 98.488.

2.2 Educación

La distribución porcentual de los alumnos con necesidades educativas especiales (NEE) derivadas de una discapacidad, matriculados en Régimen general, se corresponde a un 19,8%, los escolarizados en centros de educación especial; frente a un 80,2% que los están en centros de educación integrada.

Otros datos indispensables para conocer la situación actual, son los siguientes:

- Tasa de abandono escolar: 43,2% personas con discapacidad (en adelante PCD, frente al 25,2% de personas sin discapacidad (en adelante PSD).
- Barreras en el acceso a la formación: 69,5% de PCD, frente al 51% de PSD.
- Cursando estudios en el momento actual: 6,9% de PCD, frente al 18,5% de PSD.
- Realizando estudios superiores: 15,1% de PCD, frente al 33,2% de PSD.
- Sin estudios: 5,8% de PCD, frente al 0,5% de PSD.

Especial atención merece la *sentencia nº 1385/2017 del Tribunal Superior de Justicia de Andalucía, Sala de lo contencioso-administrativo de Málaga, sección 3ª, de 10 de julio de 2017*, que ha estimado el recurso de un alumno con discapacidad, al que se denegó su solicitud de matriculación en 2º curso de Formación Profesional Básica de Agrojardinería y Arreglos Florales por superar durante el año natural el límite de edad de 21 años establecido para permanecer cursando Ciclos Formativos de FP Básica. El citado alumno alegó que se vulneraba su derecho fundamental a la igualdad y no discriminación de acuerdo con el artículo 14 y 27 de la Constitución. La sentencia se ha dictado tras un procedimiento de derechos fundamentales, lo cual es muy positivo y además señala que el límite de 21 años no es aplicable a la FP en centros ordinarios (sino solo a la educación especial) razón por la que puede comenzarse a los 19 años y permanecer durante 4 años como señala el artículo 22 del Decreto 135/2016 de 26 de julio por el que se regulan las enseñanzas de Formación Profesional Básica en Andalucía. Sin duda, se trata de un buen precedente que contribuye a derribar las barreras existentes por razón de edad, a la que se han tenido que enfrentar hasta ahora los alumnos con discapacidad en la FP ordinaria.

2.3 Condiciones de trabajo y empleo

La situación actual en España para las PCD, respecto a sus condiciones de trabajo y desarrollo profesional, queda reflejada a través de los datos siguientes:

- El porcentaje de personas que señalan barreras para un empleo adecuado, es 90,2%.
- Tasa de pluriempleo, 2,2%.
- Tasa de autoempleo, 11,4%.
- Tasa de riesgo de pobreza o exclusión social en ocupados, 14,5%.
- Tasa de riesgo de pobreza o exclusión social general, 30,9%.
- Tasa de baja intensidad laboral, 32,9%.
- Ocupados con contratos indefinidos, 77%.
- Ocupadas con nivel de estudios secundarios, 59%.
- Ocupados con contratos a jornada completa, 83,4%.
- Contratos en el sector servicios, 80,7%.
- Permanencia de más de 3 años en el mismo empleo, 74,6%.
- El salario medio bruto anual, es 19.569,6%

2.4 Prestaciones sociales

En España, se destina un total de 18.724.489 € a prestaciones sociales por invalidez. De dicha cuantía, el 84,64% se presta en efectivo, siendo el resto en especie. Sobre el PIB, los datos apuntados representan el 1,74%.

2.5 Vida independiente[27]

El Comité de Derechos de las personas con discapacidad ha publicado una Observación General: la nº 5 dedicada *al derecho a vivir de forma independiente y a ser incluido en la Comunidad*. Este instrumento, supone un avance importantísimo a nivel internacional en la interpretación y armonización legislativa de la Convención de la ONU sobre los derechos de las personas con discapacidad. El art. 25, 2 de la ley 25/2014 de Tratados y otros Acuerdos Internacionales establece que *"En la interpretación de tratados adoptados en el ámbito de una organización internacional, se tendrá en cuenta toda norma pertinente de la organización"*. También, la mencionada Observación General nº 5 desarrolla e interpreta el art, 19 de la Convención dirigido a garantizar el derecho a la vida independiente y a ser incluido dentro de la Comunidad, con la libertad de elegir y controlar la propia vida. Se trata de uno de los artículos de ámbito más amplio y más transversal, por lo que debe tenerse en cuenta al aplicar todos los artículos de la Convención. Este derecho se refiere a todas las personas con discapacidad con independencia de sus características y circunstancias y con independencia de que hayan sido privados de su capacidad legal o de que presenten grandes necesidades de apoyo.

Se señalan dos grupos especialmente institucionalizados y vulnerables:

- Las personas con grandes necesidades de apoyo, para los cuales la financiación debe ser proporcional a las necesidades.
- Las mujeres y niñas con discapacidad, que sufren con mayor frecuencia violencia y abuso.

El citado art. 19 contiene un programa para realizar el derecho a la vida independiente (dimensión individual, derecho a emanciparse) y el derecho a ser incluido en la comunidad (dimensión social que entraña el derecho a que se desarrollen entornos inclusivos); entendiendo que *"vida independiente"* no significa vivir solo y realizar las actividades de la vida diaria sin apoyo. Significa poder elegir dónde y con quien vivir.

[27] Para profundizar sobre este tema, consultar el capítulo elaborado por ALONSO PARREÑO, M.J., correspondiente al *"Informe Olivenza 2017, sobre la situación general de la discapacidad en España"*.

Por su parte, *"estar incluido en la comunidad"* significa tener una vida social plena y tener acceso a todos los servicios ofrecidos al público y a los servicios de apoyo para personas con discapacidad que les permita participar en todas las esferas de la vida.

El Comité de Derechos de las Personas con discapacidad en su Observación General nº 5 ha reconocido avances en este derecho pero también ha destacado que todavía falta eliminar muchas barreras, como las siguientes:

- No se permite a las personas ejercer su capacidad jurídica.
- Los servicios sociales no están adaptados al modelo de vida independiente.
- Las normas y presupuestos no propician la asistencia personal y el apoyo individualizado.
- Los internamientos involuntarios.
- Falta de estrategias de desinstitucionalización.
- Actitudes negativas, estigmas y estereotipos.
- Falta de comprensión de lo que implica el comentado art. 19.
- Falta de servicios accesibles y asequibles en la comunidad (transporte, salud, escuela, vivienda, teatros, cines, edificios públicos, etc.).
- Falta de mecanismos de supervisión de la aplicación del art. 19.
- Insuficiente generalización de la discapacidad en los presupuestos generales.
- Descentralización "inapropiada" que provoca disparidades entre las posibilidades de realizar el derecho dentro de un Estado Parte.

2.6 Análisis jurisprudencial sobre la aplicación de la CDPD en España

Para finalizar, comentaremos brevemente una selección de sentencias recientes que aportan novedades sobre la aplicación de la CDPD en España.

Sentencia del Tribunal Supremo Sala 1ª, S 4-4-2017, nº 216/2017, rec. 56/2016 sobre capacidad jurídica. Esta sentencia es interesante entre otros aspectos porque fija un plazo de dos años para revisar la situación de incapacidad parcial y la necesidad de curatela.

Sentencia del Tribunal Supremo Sala 1ª, de 16 de mayo de 2017, nº 298/2017, rec. 2759/2016 sobre capacidad jurídica y curatela. En ella se establece que el juez debe oír a aquél

cuya tutela o curatela se pretenda constituir (art. 231 CC y art. 45.2 de la Ley 15/2015, de 2 de julio, de la jurisdicción voluntaria) y, de otra, porque conforme al art. 12.4 de la Convención, las salvaguardias que se adopten «asegurarán que las medidas relativas al ejercicio de la capacidad jurídica respeten los derechos, la voluntad y las preferencias de la persona.

Sentencia del Tribunal Supremo Sala 1ª Pleno, S 19-1-2017, nº 31/2017, rec. 1222/2015 en relación con la atribución de la vivienda familiar. Se solicitaba que se prologase la atribución del uso de la vivienda familiar en beneficio de un hijo con discapacidad (la vivienda era privativa del otro cónyuge y el tribunal de instancia había permitido la atribución al no dueño solo por tres años). El Tribunal Supremo dice que no cabe sostener la equiparación, en todos los ámbitos, de discapacidad con minoría de edad, que sin embargo sí que sostiene para las pensiones de alimentos. Esta sentencia tiene un pronunciamiento muy interesante:

"La condición de discapaz no deriva necesariamente de una resolución judicial dictada en juicio de modificación de la capacidad de una persona. La condición de discapaz, según el artículo 1 de la Convención de Nueva York de 2006, incluye a aquellas personas que tengan deficiencias físicas, mentales, intelectuales o sensoriales a largo plazo que, al interactuar con diversas barreras, puedan impedir su participación plena y efectiva en la sociedad, en igualdad de condiciones con las demás. Esta definición se recoge literalmente en el art. 25 del Código Penal y supone que no es precisa una declaración judicial para que puedan prestarse los apoyos necesarios a quien de hecho y no de derecho sufre alguna limitación de esta clase". (FJ2)

Sentencias del Tribunal Superior de Justicia de Madrid, Sala de lo Contencioso-administrativo, sección octava de 16 de marzo de 2017 y 13 de julio 2017 en materia de grado de dependencia y atrasos. Estas dos sentencias son importantes porque ponen en evidencia la ilegalidad de convertir en revisiones de oficio los recursos de alzada presentados por familiares de menores en situación de dependencia, situando la fecha de efectos de las prestaciones económicas reconocidas conforme a los recursos de alzada y condenando a pagar los atrasos correspondientes con intereses y costas.

Sentencia del Tribunal Supremo Sala 4ª, de 28 de junio de 2016, nº 568/2016, rec. 80/2015 sobre prestación social por reducción de jornada para cuidado de menor con cáncer u otra enfermedad grave o incurable. El Tribunal Supremo ha vuelto a confirmar, como ya hizo en la Sentencia de 15 de octubre de 2015, que es compatible esta prestación con estar escolarizado y no en un hospital.

The research has been carried out in the framework of the Erasmus+ project EC+: Reference number: 2015-1-ES01-KA203-015625

Enhancing communication: research to improve communication for people with special needs and development of ICT resources and tools.

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Co-funded by the
Erasmus+ Programme
of the European Union