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

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 syndrome	Síndrome de Angelman	Angelman-Syndrom	http://www.medigraphic.com/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/sites/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.pdf
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 disability	Discapacidad motriz	motorische Behinderung	http://www.redalyc.org/pdf/3679/367937046008.pdf
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 and Alternative Communication Systems (AAC systems)	Sistemas de Comunicación Aumentativos y Alternativos (Sistema de CAA)	unterstützte Kommunikation (UK)	https://ecplusproject.uma.es/sites/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	http://www.aeped.es/sites/default/files/documentos/sindrome_de_cornelia_de_lange.pdf

		in der psychomotorischen Entwicklung	
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 impairment[1]	Retraso del crecimiento[2]	[1](Übersetzung des engl. Terminus): Wachstumsbeeinträchtigung [2](Übersetzung des span. Terminus): Verzögerung des Wachstums	http://www.aeped.es/sites/default/files/documentos/sindrome_de_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.um

			a.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 spectrum	Espectro Óculo Aurículo Vertebra	okulo-aurikulo-vertebrales Spektrum	http://www.scielo.org.co/pdf/ceso/v28n1/v28n1a7.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	http://www.medigraphic.com/pdfs/imss/im-2011/

		Epilepsie	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&Expe rt=261494
Connective tissue disorders	Trastorno del tejido conectivo	Bindegewebserkrankung	https:// www.kleefstra syndrome.org/ what-is-kleefstra-syndrome/
Constipation	Estreñimiento	Obstipation	https:// www.orpha.net /consor/cgi-bin/ OC_Exp.php? lng=EN&Expe rt=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.kleefstra

			syndrome.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&Expert=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-	Gen histona-lisina N-	Gen der euchromatischen Histon-Lysin-N-Methyltransferase 1	https://www.orpha.net

methyltransferase 1 (*EHMT1*)	metiltransferasa 1 eucromática *(EHMT1)*	(*EHMT1*)	/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.php?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&source=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.sciencedirect.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	Desarrollo	verzögerte geistige	https://www.orpha.net/

development	mental lento	Entwicklung	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=S172

			7-897X2012000100013
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	soziale Störung	http://scielo.sld.cu/

	social		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	Autosómica	autosomal-dominant	https://

dominant	dominante		ecplusproject.uma.es/sites/default/files/guide-eng.pdf
Chromosome 1	Cromosoma 1	Chromosom 1	https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf
Comparative genomic hybridization	Hibridación genómica comparativa	vergleichende Genom-Hybridisierung (CGH)	https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf
Deletion	Deleción	Deletion	https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf
Dental malocclusion /Dental oclusal disharmony	Maloclusión dental	dentale Malokklusion	https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf
Dolichocephaly	Dolicocefalia	Dolichozephalie	https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf
Facial dimorphism	Dimorfismo facial	faziale Dysmorphie	https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf
Hypotony	Hipotonía	Hypotonie	https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf
Inbred	Consanguíneo	blutsverwandt	https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf
Microcephaly	Microcefalia	Mikrozephalie	https://ecplusproject.uma.es/sites/default/files/

			guide-eng.pdf
Moderate deficit	Déficit moderado	leichte Behinderung	https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf
Novo gene mutation	Mutación de novo	Neumutation	https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf
Phenotype	Fenotipo	Phänotyp	http://www.analesdepediatria.org/es-sndrome-microdelecin-22q11-manifestaciones-cardiorrespiratorias-articulo-S1695403312000951
Retrognathia	Retrognatia	Retrognathie	https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf
Severe speech delay	Retardo grave (en el lenguajeo)	schwere verzögerte Sprachentwicklung	https://ecplusproject.uma.es/sites/default/files/guide-eng.pdf

10. Mowat-Wilson syndrome

ENGLISH	**SPANISH**	**GERMAN**	**REFERENCES**
Agenesis of corpus callosum	Agenesia del cuerpo calloso	Agenesie des Corpus callosum	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152
Arterial duct	Canal arterial	arterierer Kanal	https://www.ncbi.nlm.nih.gov/medgen/4415
Atypical phenotype	Fenotipo atípico	atypische Symptomatik	https://www.orpha.net/consor/cgi-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 psychomotor development	Retraso en el desarrollo psicomotor	verzögerte psychomotorische Entwicklung	https://www.orpha.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	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2152

		(Physiotherapie, psychomotorische und Sprachtherapie) sollte daher so früh wie möglich begonnen werden	
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/cgi-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	Extreñimiento	schwere Obstipation	http://www.ivami.com/en/

constipation	grave		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-pharmacogenetics/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	Infradiagnostic ado	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/articles/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	Uñas	dysplastische	https://ojrd.biomedcentral.com/

nails	displásicas	Zehennägel	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/10.1186/1750-1172-3-14
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/articles/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
synGlobal developmental delay	Retraso global del desarrollo	allgemeine Entwicklungsverzögerung	https://www.orpha.net/consor/cgi-bin/OC_Exp.php?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 syndrome	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 syndrome	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 helices	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 heterocigotas	heterozygote Neumutationen	https://www.orpha.net/consor/cgi-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 deficit	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 syndrome	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/cgi-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	allgemeiner Bedarf an sanitären Diensten	http://www.who.int/

	generales		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.autismparentingmagazine.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 acute and chronic illness	Tratamiento de enfermedades agudas y crónicas	Behandlung akuter und chronischer Erkrankungen	http://www.who.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.eyeglossary.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	Adquisición del	Spracherwerb	https://

acquisition	lenguaje		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.php?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 (electroencephal ogram)	Electroencefal ograma	Elektroenzephalogra mm (EEG)	https://www.siis.net/ documentos/ficha/217518.pdf
Electroclinical triad	Tríada electroclínica	elelektroklinische Symptomen-Trias	http:// www.revistachilenadeepilepsia .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
Hyperbilirubine mia	Hiperbilirrubin emia	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