Helen Leonard

List of Publications by Citations

314 papers	10,795 citations	53 h-index	88 g-index
336 ext. papers	12,735 ext. citations	3.9 avg, IF	6.16 L-index

#	Paper	IF	Citations
314	Rett syndrome: revised diagnostic criteria and nomenclature. *Annals of Neurology*, **2010**, 68, 944-50	9.4	804
313	The epidemiology of mental retardation: challenges and opportunities in the new millennium. *Mental Retardation and Developmental Disabilities Research Reviews*, **2002**, 8, 117-34		428
312	Mutations of CDKL5 cause a severe neurodevelopmental disorder with infantile spasms and mental retardation. *American Journal of Human Genetics*, **2004**, 75, 1079-93	11	352
311	Rett syndrome in Australia: a review of the epidemiology. *Journal of Pediatrics*, **2006**, 148, 347-52	3.6	241
310	Intellectual disability co-occurring with schizophrenia and other psychiatric illness: population-based study. *British Journal of Psychiatry*, **2008**, 193, 364-72	5.4	226
309	Long-read sequence analysis of the MECP2 gene in Rett syndrome patients: correlation of disease severity with mutation type and location. *Human Molecular Genetics*, **2000**, 9, 1119-29	5.6	196
308	Investigating genotype-phenotype relationships in Rett syndrome using an international data set. *Neurology*, **2008**, 70, 868-75	6.5	180
307	The CDKL5 disorder is an independent clinical entity associated with early-onset encephalopathy. *European Journal of Human Genetics*, **2013**, 21, 266-73	5.3	161
306	Association of Genetic and Environmental Factors With Autism in a 5-Country Cohort. *JAMA Psychiatry*, **2019**, 76, 1035-1043	14.5	151
305	The increasing prevalence of reported diagnoses of childhood psychiatric disorders: a descriptive multinational comparison. *European Child and Adolescent Psychiatry*, **2015**, 24, 173-83	5.5	147
304	Autism risk associated with parental age and with increasing difference in age between the parents. *Molecular Psychiatry*, **2016**, 21, 693-700	15.1	129
303	Guidelines for reporting clinical features in cases with MECP2 mutations. *Brain and Development*, **2001**, 23, 208-11	2.2	121
302	Clinical and biological progress over 50 years in Rett syndrome. *Nature Reviews Neurology*, **2017**, 13, 37-51	15	120
301	Autism spectrum disorders in young children: effect of changes in diagnostic practices. *International Journal of Epidemiology*, **2009**, 38, 1245-54	7.8	101
300	Sleep problems in Rett syndrome. *Brain and Development*, **2007**, 29, 609-16	2.2	91
299	Survival of infants born with Down's syndrome: 1980-96. *Paediatric and Perinatal Epidemiology*, **2000**, 14, 163-71	2.7	91
298	Trends in the diagnosis of Rett syndrome in Australia. *Pediatric Research*, **2011**, 70, 313-9	3.2	90

297	Describing the phenotype in Rett syndrome using a population database. *Archives of Disease in Childhood*, **2003**, 88, 38-43	2.2	88
296	Rare childhood diseases: how should we respond?. *Archives of Disease in Childhood*, **2008**, 93, 1071-4	2.2	85
295	Effects of MECP2 mutation type, location and X-inactivation in modulating Rett syndrome phenotype. *American Journal of Medical Genetics Part A*, **2003**, 118A, 103-14		84
294	Autism and intellectual disability are differentially related to sociodemographic background at birth. *PLoS ONE*, **2011**, 6, e17875	3.7	82
293	Increasing prevalence of neonatal withdrawal syndrome: population study of maternal factors and child protection involvement. *Pediatrics*, **2009**, 123, e614-21	7.4	81
292	Predictors of seizure onset in Rett syndrome. *Journal of Pediatrics*, **2006**, 149, 542-7	3.6	79
291	Association of sociodemographic characteristics of children with intellectual disability in Western Australia. *Social Science and Medicine*, **2005**, 60, 1499-513	5.1	79
290	Maternal conditions and perinatal characteristics associated with autism spectrum disorder and intellectual disability. *PLoS ONE*, **2013**, 8, e50963	3.7	78
289	Prevalence of intellectual disability in Western Australia. *Paediatric and Perinatal Epidemiology*, **2003**, 17, 58-67	2.7	76
288	Young adults with intellectual disability transitioning from school to post-school: a literature review framed within the ICF. *Disability and Rehabilitation*, **2012**, 34, 1747-64	2.4	75
287	The impact of having a sibling with an intellectual disability: parental perspectives in two disorders. *Journal of Intellectual Disability Research*, **2008**, 52, 216-29	3.2	75
286	Early progressive encephalopathy in boys and MECP2 mutations. *Neurology*, **2006**, 67, 164-6	6.5	75
285	Physical and mental health in mothers of children with Down syndrome. *Journal of Pediatrics*, **2008**, 153, 320-6	3.6	73
284	Is the girl with Rett syndrome normal at birth?. *Developmental Medicine and Child Neurology*, **1998**, 40, 115-21	3.3	73
283	Twenty years of surveillance in Rett syndrome: what does this tell us?. *Orphanet Journal of Rare Diseases*, **2014**, 9, 87	4.2	72
282	Correlation between clinical severity in patients with Rett syndrome with a p.R168X or p.T158M MECP2 mutation, and the direction and degree of skewing of X-chromosome inactivation. *Journal of Medical Genetics*, **2007**, 44, 148-52	5.8	72
281	Association of gestational age and growth measures at birth with infection-related admissions to hospital throughout childhood: a population-based, data-linkage study from Western Australia. *Lancet Infectious Diseases, The*, **2016**, 16, 952-61	25.5	70
280	Seizures in Rett syndrome: an overview from a one-year calendar study. *European Journal of Paediatric Neurology*, **2007**, 11, 310-7	3.8	68

279	Patients with the R133C mutation: is their phenotype different from patients with Rett syndrome with other mutations?. *Journal of Medical Genetics*, **2003**, 40, e52	5.8	67
278	Australian children living with rare diseases: experiences of diagnosis and perceived consequences of diagnostic delays. *Orphanet Journal of Rare Diseases*, **2017**, 12, 68	4.2	66
277	Refining the phenotype of common mutations in Rett syndrome. *Journal of Medical Genetics*, **2004**, 41, 25-30	5.8	65
276	Epilepsy and mental retardation limited to females with PCDH19 mutations can present de novo or in single generation families. *Journal of Medical Genetics*, **2010**, 47, 211-6	5.8	64
275	Population-Based Prevalence of Intellectual Disability and Autism Spectrum Disorders in Western Australia: A Comparison With Previous Estimates. *Medicine (United States)*, **2016**, 95, e3737	1.8	64
274	Maltreatment Risk Among Children With Disabilities. *Pediatrics*, **2017**, 139,	7.4	62
273	Linking MECP2 and pain sensitivity: the example of Rett syndrome. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1197-205	2.5	62
272	Occurrence of Rett syndrome in boys. *Journal of Child Neurology*, **2001**, 16, 333-8	2.5	62
271	Cyclin-Dependent Kinase-Like 5 Deficiency Disorder: Clinical Review. *Pediatric Neurology*, **2019**, 97, 18-25	2.9	60
270	Early determinants of fractures in Rett syndrome. *Pediatrics*, **2008**, 121, 540-6	7.4	60
269	The trajectories of sleep disturbances in Rett syndrome. *Journal of Sleep Research*, **2015**, 24, 223-33	5.8	59
268	The common BDNF polymorphism may be a modifier of disease severity in Rett syndrome. *Neurology*, **2009**, 72, 1242-7	6.5	59
267	Relation between intrauterine growth and subsequent intellectual disability in a ten-year population cohort of children in Western Australia. *American Journal of Epidemiology*, **2008**, 167, 103-11	3.8	59
266	IDEA (Intellectual Disability Exploring Answers): a population-based database for intellectual disability in Western Australia. *Annals of Human Biology*, **2005**, 32, 237-43	1.7	59
265	Physical and mental health of mothers caring for a child with Rett syndrome. *Pediatrics*, **2006**, 118, e1152-64	7.4	57
264	Family functioning in families with a child with Down syndrome: a mixed methods approach. *Journal of Intellectual Disability Research*, **2012**, 56, 961-73	3.2	55
263	To Feel Belonged: The Voices of Children and Youth with Disabilities on the Meaning of Wellbeing. *Child Indicators Research*, **2012**, 5, 375-391	1.9	54
262	Maternal health in pregnancy and intellectual disability in the offspring: a population-based study. *Annals of Epidemiology*, **2006**, 16, 448-54	6.4	54

261	Health conditions and their impact among adolescents and young adults with Down syndrome. *PLoS ONE*, **2014**, 9, e96868	3.7	52
260	Seizure variables and their relationship to genotype and functional abilities in the CDKL5 disorder. *Neurology*, **2016**, 87, 2206-2213	6.5	51
259	Prevalence and onset of comorbidities in the CDKL5 disorder differ from Rett syndrome. *Orphanet Journal of Rare Diseases*, **2016**, 11, 39	4.2	51
258	A comparison of autism prevalence trends in Denmark and Western Australia. *Journal of Autism and Developmental Disorders*, **2011**, 41, 1601-8	4.6	51
257	Updating the profile of C-terminal MECP2 deletions in Rett syndrome. *Journal of Medical Genetics*, **2010**, 47, 242-8	5.8	51
256	Unpacking the complex nature of the autism epidemic. *Research in Autism Spectrum Disorders*, **2010** , 4, 548-554	3	51
255	Predictors of scoliosis in Rett syndrome. *Journal of Child Neurology*, **2006**, 21, 809-13	2.5	51
254	Guidelines for management of scoliosis in Rett syndrome patients based on expert consensus and clinical evidence. *Spine*, **2009**, 34, E607-17	3.3	50
253	Risk of stillbirth, preterm delivery, and fetal growth restriction following exposure in a previous birth: systematic review and meta-analysis. *BJOG: an International Journal of Obstetrics and Gynaecology*, **2018**, 125, 183-192	3.7	49
252	Prolonged QT interval in Rett syndrome. *Archives of Disease in Childhood*, **1999**, 80, 470-2	2.2	49
251	The prevalence of mental health disorders and symptoms in children and adolescents with cerebral palsy: a systematic review and meta-analysis. *Developmental Medicine and Child Neurology*, **2018**, 60, 30-38	3.3	48
250	The diagnosis of autism in a female: could it be Rett syndrome?. *European Journal of Pediatrics*, **2008**, 167, 661-9	4.1	48
249	There is variability in the attainment of developmental milestones in the CDKL5 disorder. *Journal of Neurodevelopmental Disorders*, **2015**, 7, 2	4.6	47
248	Using a large international sample to investigate epilepsy in Rett syndrome. *Developmental Medicine and Child Neurology*, **2013**, 55, 553-8	3.3	47
247	Gross motor profile in rett syndrome as determined by video analysis. *Neuropediatrics*, **2008**, 39, 205-10	1.6	47
246	Leisure participation for school-aged children with Down syndrome. *Disability and Rehabilitation*, **2011**, 33, 1880-9	2.4	46
245	Maternal mental health and risk of child protection involvement: mental health diagnoses associated with increased risk. *Journal of Epidemiology and Community Health*, **2015**, 69, 1175-83	5.1	45
244	The association between behavior and genotype in Rett syndrome using the Australian Rett Syndrome Database. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2006**, 141B, 177-83	3.5	45

243	Functional status of school-aged children with Down syndrome. *Journal of Paediatrics and Child Health*, **2002**, 38, 160-5	1.3	45
242	Hospital admissions in children with down syndrome: experience of a population-based cohort followed from birth. *PLoS ONE*, **2013**, 8, e70401	3.7	44
241	Functional status, medical impairments, and rehabilitation resources in 84 females with Rett syndrome: a snapshot across the world from the parental perspective. *Disability and Rehabilitation*, **2001**, 23, 107-17	2.4	44
240	Young People with Intellectual Disability Transitioning to Adulthood: Do Behaviour Trajectories Differ in Those with and without Down Syndrome?. *PLoS ONE*, **2016**, 11, e0157667	3.7	44
239	Variation over time in medical conditions and health service utilization of children with Down syndrome. *Journal of Pediatrics*, **2011**, 158, 194-200.e1	3.6	43
238	Characteristics of non-Aboriginal and Aboriginal children and families with substantiated child maltreatment: a population-based study. *International Journal of Epidemiology*, **2010**, 39, 921-8	7.8	42
237	Stereotypical hand movements in 144 subjects with Rett syndrome from the population-based Australian database. *Movement Disorders*, **2010**, 25, 282-8	7	42
236	Validating the Rett Syndrome Gross Motor Scale. *PLoS ONE*, **2016**, 11, e0147555	3.7	42
235	Intellectual disability: population-based estimates of the proportion attributable to maternal alcohol use disorder during pregnancy. *Developmental Medicine and Child Neurology*, **2013**, 55, 271-7	3.3	41
234	Rett syndrome: randomized controlled trial of L-carnitine. *Journal of Child Neurology*, **1999**, 14, 162-7	2.5	41
233	The Natural History of Scoliosis in Females With Rett Syndrome. *Spine*, **2016**, 41, 856-63	3.3	40
232	Delineation of large deletions of the MECP2 gene in Rett syndrome patients, including a familial case with a male proband. *European Journal of Human Genetics*, **2007**, 15, 1218-29	5.3	40
231	InterRett, a model for international data collection in a rare genetic disorder. *Research in Autism Spectrum Disorders*, **2009**, 3, 639-639	3	39
230	Transition to adulthood for young people with intellectual disability: the experiences of their families. *European Child and Adolescent Psychiatry*, **2016**, 25, 1369-1381	5.5	39
229	Improved Survival in Down Syndrome over the Last 60 Years and the Impact of Perinatal Factors in Recent Decades. *Journal of Pediatrics*, **2016**, 169, 214-20.e1	3.6	38
228	A population-based approach to the investigation of osteopenia in Rett syndrome. *Developmental Medicine and Child Neurology*, **1999**, 41, 323-8	3.3	38
227	Assessment and management of nutrition and growth in Rett syndrome. *Journal of Pediatric Gastroenterology and Nutrition*, **2013**, 57, 451-60	2.8	37
226	Level of purposeful hand function as a marker of clinical severity in Rett syndrome. *Developmental Medicine and Child Neurology*, **2010**, 52, 817-23	3.3	37

225	NTNG1 mutations are a rare cause of Rett syndrome. *American Journal of Medical Genetics, Part A*, **2006**, 140, 691-4	2.5	37
224	Functional abilities in children and adults with the CDKL5 disorder. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2860-2869	2.5	36
223	Conceptualizing a quality of life framework for girls with Rett syndrome using qualitative methods. *American Journal of Medical Genetics, Part A*, **2016**, 170, 645-53	2.5	36
222	Gastrointestinal dysmotility in Rett syndrome. *Journal of Pediatric Gastroenterology and Nutrition*, **2014**, 58, 237-44	2.8	36
221	Bone mineral content and density in Rett syndrome and their contributing factors. *Pediatric Research*, **2011**, 69, 293-8	3.2	36
220	Environmental enrichment intervention for Rett syndrome: an individually randomised stepped wedge trial. *Orphanet Journal of Rare Diseases*, **2018**, 13, 3	4.2	35
219	"It's not what you were expecting, but it's still a beautiful journey": the experience of mothers of children with Down syndrome. *Disability and Rehabilitation*, **2012**, 34, 1501-10	2.4	35
218	Survival with Rett syndrome: comparing Rett's original sample with data from the Australian Rett Syndrome Database. *Developmental Medicine and Child Neurology*, **2010**, 52, 962-5	3.3	35
217	Feeding experiences and growth status in a Rett syndrome population. *Journal of Pediatric Gastroenterology and Nutrition*, **2007**, 45, 582-90	2.8	35
216	Co-occurrence of birth defects and intellectual disability. *Paediatric and Perinatal Epidemiology*, **2007**, 21, 65-75	2.7	34
215	ViPAR: a software platform for the Virtual Pooling and Analysis of Research Data. *International Journal of Epidemiology*, **2016**, 45, 408-416	7.8	33
214	Altered attainment of developmental milestones influences the age of diagnosis of rett syndrome. *Journal of Child Neurology*, **2011**, 26, 980-7	2.5	33
213	Sleep dysfunction in Rett syndrome: lack of age related decrease in sleep duration. *Brain and Development*, **2001**, 23 Suppl 1, S101-3	2.2	33
212	Clinical Guidelines for Management of Bone Health in Rett Syndrome Based on Expert Consensus and Available Evidence. *PLoS ONE*, **2016**, 11, e0146824	3.7	33
211	The prevalence and incidence of Rett syndrome in Australia. *European Child and Adolescent Psychiatry*, **1997**, 6 Suppl 1, 8-10	5.5	33
210	Expanding the clinical picture of the MECP2 Duplication syndrome. *Clinical Genetics*, **2017**, 91, 557-563	4	32
209	Role of public and private funding in the rising caesarean section rate: a cohort study. *BMJ Open*, **2013**, 3,	3	32
208	"I have a good life": the meaning of well-being from the perspective of young adults with Down syndrome. *Disability and Rehabilitation*, **2014**, 36, 1290-8	2.4	31

207	The experiences of mothers of young adults with an intellectual disability transitioning from secondary school to adult life. *Journal of Intellectual and Developmental Disability*, **2013**, 38, 149-62	1.9	31
206	Development of a video-based evaluation tool in Rett syndrome. *Journal of Autism and Developmental Disorders*, **2007**, 37, 1636-46	4.6	31
205	How can the Internet help parents of children with rare neurologic disorders?. *Journal of Child Neurology*, **2004**, 19, 902-7	2.5	31
204	Medium-term open label trial of L-carnitine in Rett syndrome. *Brain and Development*, **2001**, 23 Suppl 1, S85-9	2.2	31
203	Caesarean section and risk of autism across gestational age: a multi-national cohort study of 5 million births. *International Journal of Epidemiology*, **2017**, 46, 429-439	7.8	31
202	Use of the ketogenic diet to manage refractory epilepsy in CDKL5 disorder: Experience of >100 patients. *Epilepsia*, **2017**, 58, 1415-1422	6.4	30
201	Change in gross motor abilities of girls and women with rett syndrome over a 3- to 4-year period. *Journal of Child Neurology*, **2011**, 26, 1237-45	2.5	30
200	InterRett and RettBASE: International Rett Syndrome Association databases for Rett syndrome. *Journal of Child Neurology*, **2003**, 18, 709-13	2.5	30
199	Genotype and early development in Rett syndrome: the value of international data. *Brain and Development*, **2005**, 27 Suppl 1, S59-S68	2.2	30
198	Impacts of caring for a child with the CDKL5 disorder on parental wellbeing and family quality of life. *Orphanet Journal of Rare Diseases*, **2017**, 12, 16	4.2	29
197	Early development and regression in Rett syndrome. *Clinical Genetics*, **2013**, 84, 572-6	4	29
196	Valproate and risk of fracture in Rett syndrome. *Archives of Disease in Childhood*, **2010**, 95, 444-8	2.2	29
195	Association of birth outcomes and maternal, school, and neighborhood characteristics with subsequent numeracy achievement. *American Journal of Epidemiology*, **2008**, 168, 21-9	3.8	28
194	p.R270X MECP2 mutation and mortality in Rett syndrome. *European Journal of Human Genetics*, **2005**, 13, 1235-8	5.3	28
193	variants: Improving our understanding of a rare neurologic disorder. *Neurology: Genetics*, **2017**, 3, e200	3.8	27
192	Hospitalisation rates for children with intellectual disability or autism born in Western Australia 1983-1999: a population-based cohort study. *BMJ Open*, **2013**, 3,	3	27
191	Monitoring child abuse and neglect at a population level: patterns of hospital admissions for maltreatment and assault. *Child Abuse and Neglect*, **2010**, 34, 823-32	4.3	27
190	Atypical presentations and specific genotypes are associated with a delay in diagnosis in females with Rett syndrome. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 2535-42	2.5	27

189	Hospitalisations from birth to 5 years in a population cohort of Western Australian children with intellectual disability. *Archives of Disease in Childhood*, **2005**, 90, 1243-8	2.2	27
188	Association of Gestational Age at Birth with Reasons for Subsequent Hospitalisation: 18 Years of Follow-Up in a Western Australian Population Study. *PLoS ONE*, **2015**, 10, e0130535	3.7	27
187	Direct health care costs of children and adolescents with Down syndrome. *Journal of Pediatrics*, **2011**, 159, 541-5	3.6	26
186	Siblings of children with disabilities: challenges and opportunities. *Acta Paediatrica, International Journal of Paediatrics*, **2009**, 98, 23-4	3.1	26
185	Prevalence estimates of mental health problems in children and adolescents with intellectual disability: A systematic review and meta-analysis. *Australian and New Zealand Journal of Psychiatry*, **2020**, 54, 970-984	2.6	25
184	Functioning and post-school transition outcomes for young people with Down syndrome. *Child: Care, Health and Development*, **2013**, 39, 789-800	2.8	25
183	Exploring quality of life of children with cerebral palsy and intellectual disability: What are the important domains of life?. *Child: Care, Health and Development*, **2017**, 43, 854-860	2.8	25
182	The phenotype associated with a large deletion on MECP2. *European Journal of Human Genetics*, **2012**, 20, 921-7	5.3	25
181	Qualitative Analysis of Parental Observations on Quality of Life in Australian Children with Down Syndrome. *Journal of Developmental and Behavioral Pediatrics*, **2017**, 38, 161-168	2.4	24
180	Aspects of speech-language abilities are influenced by MECP2 mutation type in girls with Rett syndrome. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 354-62	2.5	24
179	Medical aspects of school-aged children with Down syndrome. *Developmental Medicine and Child Neurology*, **1999**, 41, 683-8	3.3	24
178	Association between interpregnancy interval and adverse birth outcomes in women with a previous stillbirth: an international cohort study. *Lancet, The*, **2019**, 393, 1527-1535	40	24
177	Changes in risk factors for preterm birth in Western Australia 1984-2006. *BJOG: an International Journal of Obstetrics and Gynaecology*, **2013**, 120, 1051-60	3.7	23
176	Lost in translation: translational interference from a recurrent mutation in exon 1 of MECP2. *Journal of Medical Genetics*, **2006**, 43, 470-7	5.8	23
175	Intellectual disability in Western Australia. *Journal of Paediatrics and Child Health*, **2000**, 36, 213-5	1.3	23
174	Determinants of sleep disturbances in Rett syndrome: Novel findings in relation to genotype. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2292-300	2.5	23
173	Vagus nerve stimulation for the treatment of refractory epilepsy in the CDKL5 Deficiency Disorder. *Epilepsy Research*, **2018**, 146, 36-40	3	22
172	Rates and types of hospitalisations for children who have subsequent contact with the child protection system: a population based case-control study. *Journal of Epidemiology and Community Health*, **2010**, 64, 784-8	5.1	22

171	Psychometric properties of the Quality of Life Inventory-Disability (QI-Disability) measure. *Quality of Life Research*, **2019**, 28, 783-794	3.7	22
170	Quantification of walking-based physical activity and sedentary time in individuals with Rett syndrome. *Developmental Medicine and Child Neurology*, **2017**, 59, 605-611	3.3	21
169	Rett syndrome: establishing a novel outcome measure for walking activity in an era of clinical trials for rare disorders. *Disability and Rehabilitation*, **2015**, 37, 1992-6	2.4	21
168	Rare disease: a national survey of paediatriciansSexperiences and needs. *BMJ Paediatrics Open*, **2017**, 1, e000172	2.4	21
167	Factors associated with dental admissions for children aged under 5 years in Western Australia. *Archives of Disease in Childhood*, **2009**, 94, 517-23	2.2	21
166	The International Collaboration for Autism Registry Epidemiology (iCARE): multinational registry-based investigations of autism risk factors and trends. *Journal of Autism and Developmental Disorders*, **2013**, 43, 2650-63	4.6	20
165	Autonomic breathing abnormalities in Rett syndrome: caregiver perspectives in an international database study. *Journal of Neurodevelopmental Disorders*, **2017**, 9, 15	4.6	20
164	Relationship between family quality of life and day occupations of young people with Down syndrome. *Social Psychiatry and Psychiatric Epidemiology*, **2014**, 49, 1455-65	4.5	20
163	The impact of single gene and chromosomal disorders on hospital admissions of children and adolescents: a population-based study. *Public Health Genomics*, **2011**, 14, 153-61	1.9	20
162	Influence of the environment on participation in social roles for young adults with down syndrome. *PLoS ONE*, **2014**, 9, e108413	3.7	20
161	Parental perspectives on the communication abilities of their daughters with Rett syndrome. *Developmental Neurorehabilitation*, **2016**, 19, 17-25	1.8	19
160	Use of health services in the last year of life and cause of death in people with intellectual disability: a retrospective matched cohort study. *BMJ Open*, **2018**, 8, e020268	3	19
159	Sleep disturbances in Rett syndrome: Impact and management including use of sleep hygiene practices. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1569-1577	2.5	19
158	Experience of gastrostomy using a quality care framework: the example of rett syndrome. *Medicine (United States)*, **2014**, 93, e328	1.8	19
157	Hospital admissions and gestational age at birth: 18 years of follow up in Western Australia. *Paediatric and Perinatal Epidemiology*, **2014**, 28, 536-44	2.7	19
156	Evaluation of the processes of family-centred care for young children with intellectual disability in Western Australia. *Child: Care, Health and Development*, **2010**, 36, 709-18	2.8	19
155	Skeletal abnormalities in Rett syndrome: increasing evidence for dysmorphogenetic defects. *American Journal of Medical Genetics Part A*, **1995**, 58, 282-5		19
154	Surgical fusion of early onset severe scoliosis increases survival in Rett syndrome: a cohort study. *Developmental Medicine and Child Neurology*, **2016**, 58, 632-8	3.3	19

153	Severity Assessment in CDKL5 Deficiency Disorder. *Pediatric Neurology*, **2019**, 97, 38-42	2.9	18
152	Brief Report: Burden of Care in Mothers of Children with Autism Spectrum Disorder or Intellectual Disability. *Journal of Autism and Developmental Disorders*, **2016**, 46, 1103-9	4.6	18
151	Metacarpophalangeal pattern profile and bone age in Rett syndrome: further radiological clues to the diagnosis. *American Journal of Medical Genetics Part A*, **1999**, 83, 88-95		18
150	Very Early Identification and Intervention for Infants at Risk of Neurodevelopmental Disorders: A Transdiagnostic Approach. *Child Development Perspectives*, **2019**, 13, 97-103	5.5	17
149	Recurrence Risk of Autism in Siblings and Cousins: A Multinational, Population-Based Study. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2019**, 58, 866-875	7.2	17
148	Systematic Review and Meta-analysis: Mental Health in Children With Neurogenetic Disorders Associated With Intellectual Disability. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2020**, 59, 1036-1048	7.2	17
147	Onset of maternal psychiatric disorders after the birth of a child with intellectual disability: a retrospective cohort study. *Journal of Psychiatric Research*, **2015**, 61, 223-30	5.2	17
146	Friedreich's ataxia presenting after cardiac transplantation. *Archives of Disease in Childhood*, **2001**, 84, 167-8	2.2	17
145	Using the Internet to pilot a questionnaire on childhood disability in Rett syndrome. *Child: Care, Health and Development*, **2001**, 27, 535-43	2.8	16
144	Validation of intellectual disability coding through hospital morbidity records using an intellectual disability population-based database in Western Australia. *BMJ Open*, **2018**, 8, e019113	3	15
143	Trends in pre-existing mental health disorders among parents of infants born in Western Australia from 1990 to 2005. *Medical Journal of Australia*, **2013**, 198, 485-8	4	15
142	Longitudinal hand function in Rett syndrome. *Journal of Child Neurology*, **2011**, 26, 334-40	2.5	15
141	A Population-Based Matched-Sibling Analysis Estimating the Associations Between First Interpregnancy Interval and Birth Outcomes. *American Journal of Epidemiology*, **2019**, 188, 9-16	3.8	15
140	Longitudinal effects of caregiving on parental well-being: the example of Rett syndrome, a severe neurological disorder. *European Child and Adolescent Psychiatry*, **2019**, 28, 505-520	5.5	15
139	Apgar score and risk of autism. *European Journal of Epidemiology*, **2019**, 34, 105-114	12.1	15
138	Parent-observed thematic data on quality of life in children with autism spectrum disorder. *Autism*, **2019**, 23, 71-80	6.6	15
137	Intellectual Disability in Children Conceived Using Assisted Reproductive Technology. *Pediatrics*, **2018**, 142,	7.4	15
136	Cannabis for refractory epilepsy in children: A review focusing on CDKL5 Deficiency Disorder. *Epilepsy Research*, **2019**, 151, 31-39	3	14

135	Evolving Trends of Gastrostomy Insertion Within a Pediatric Population. *Journal of Pediatric Gastroenterology and Nutrition*, **2018**, 67, e89-e94	2.8	14
134	Patterns of depressive symptoms and social relating behaviors differ over time from other behavioral domains for young people with Down syndrome. *Medicine (United States)*, **2015**, 94, e710	1.8	14
133	Building the repertoire of measures of walking in Rett syndrome. *Disability and Rehabilitation*, **2017**, 39, 1926-1931	2.4	14
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