# Helen Leonard

# List of Publications by Year in Descending Order

Source: https://exaly.com/author-pdf/672432/helen-leonard-publications-by-year.pdf

Version: 2024-04-10

This document has been generated based on the publications and citations recorded by exaly.com. For the latest version of this publication list, visit the link given above.

The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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

#	Paper	IF	Citations
314	Child protection involvement of children of mothers with intellectual disability <i>Child Abuse and Neglect</i> , <b>2022</b> , 126, 105515	4.3	O
313	Epidemiology of Hospital Admissions for Craniosynostosis in Australia: A Population-Based Study <i>Cleft Palate-Craniofacial Journal</i> , <b>2022</b> , 10556656221074215	1.9	0
312	Modifiable child and caregiver factors that influence community participation among children with Down syndrome <i>Disability and Rehabilitation</i> , <b>2022</b> , 44, 600-607	2.4	2
311	The Lived Experience of ParentsSReceiving the Diagnosis of CDKL5 Deficiency Disorder for Their Child <i>Journal of Child Neurology</i> , <b>2022</b> , 8830738221076285	2.5	
310	Daytime sleepiness and emotional and behavioral disturbances in Prader-Willi syndrome <i>European Journal of Pediatrics</i> , <b>2022</b> , 1	4.1	
309	Improving clinical trial readiness to accelerate development of new therapeutics for Rett syndrome <i>Orphanet Journal of Rare Diseases</i> , <b>2022</b> , 17, 108	4.2	0
308	A brief history of MECP2 duplication syndrome: 20-years of clinical understanding <i>Orphanet Journal of Rare Diseases</i> , <b>2022</b> , 17, 131	4.2	O
307	Initial Validation and Reliability of the CDKL5 Deficiency Disorder Hand Function Scale (CDD-Hand) <i>Journal of Child Neurology</i> , <b>2022</b> , 8830738221091044	2.5	0
306	Negative impact of insomnia and daytime sleepiness on quality of life in individuals with the cyclin-dependent kinase-like 5 deficiency disorder <i>Journal of Sleep Research</i> , <b>2022</b> , e13600	5.8	0
305	Medical Comorbidities in MECP2 Duplication Syndrome: Results from the International MECP2 Duplication Database. <i>Children</i> , <b>2022</b> , 9, 633	2.8	1
304	Parent and therapist perspectives on "uptime" activities and participation in Rett syndrome. <i>Disability and Rehabilitation</i> , <b>2021</b> , 1-8	2.4	
303	Epidemiology of Rare Craniofacial Anomalies: Retrospective Western Australian Population Data Linkage Study. <i>Journal of Pediatrics</i> , <b>2021</b> ,	3.6	2
302	Determinants of quality of life in Rett syndrome: new findings on associations with genotype. <i>Journal of Medical Genetics</i> , <b>2021</b> , 58, 637-644	5.8	4
301	Oral health care and service utilisation in individuals with Rett syndrome: an international cross-sectional study. <i>Journal of Intellectual Disability Research</i> , <b>2021</b> , 65, 561-576	3.2	2
300	Comorbidities and quality of life in children with intellectual disability. <i>Child: Care, Health and Development</i> , <b>2021</b> , 47, 654-666	2.8	1
299	Determinants of sleep problems in children with intellectual disability. <i>Journal of Sleep Research</i> , <b>2021</b> , 30, e13361	5.8	2
298	The effect of functioning on Quality of Life Inventory-Disability measured quality of life is not mediated or moderated by parental psychological distress. <i>Quality of Life Research</i> , <b>2021</b> , 30, 2875-2885	<del>3</del> .7	O

### (2020-2021)

297	Oral parafunction and bruxism in Rett syndrome and associated factors: An observational study. Oral Diseases, <b>2021</b> ,	3.5	1	
296	Factors associated with dental hospitalisations in children with intellectual disability or autism spectrum disorder: a Western Australian population-based retrospective cohort study. <i>Disability and Rehabilitation</i> , <b>2021</b> , 1-9	2.4	1	
295	Exploring genotype-phenotype relationships in the CDKL5 deficiency disorder using an international dataset. <i>Clinical Genetics</i> , <b>2021</b> , 99, 157-165	4	5	
294	Exploring quality of life in individuals with a severe developmental and epileptic encephalopathy, CDKL5 Deficiency Disorder. <i>Epilepsy Research</i> , <b>2021</b> , 169, 106521	3	7	
293	Functioning, participation, and quality of life in children with intellectual disability: an observational study. <i>Developmental Medicine and Child Neurology</i> , <b>2021</b> , 63, 89-96	3.3	12	
292	Optimal interpregnancy interval in autism spectrum disorder: A multi-national study of a modifiable risk factor. <i>Autism Research</i> , <b>2021</b> , 14, 2432-2443	5.1	0	
291	Description of Total Population Hospital Admissions for Treacher Collins Syndrome in Australia. <i>Cleft Palate-Craniofacial Journal</i> , <b>2021</b> , 10556656211036618	1.9	О	
290	Content Validation of Clinician-Reported Items for a Severity Measure for CDKL5 Deficiency Disorder. <i>Journal of Child Neurology</i> , <b>2021</b> , 36, 998-1006	2.5	2	
289	Associations Between Hyperphagia, Symptoms of Sleep Breathing Disorder, Behaviour Difficulties and Caregiver Well-Being in Prader-Willi Syndrome: A Preliminary Study. <i>Journal of Autism and Developmental Disorders</i> , <b>2021</b> , 1	4.6		
288	The perceived effects of cannabis products in the management of seizures in CDKL5 Deficiency Disorder. <i>Epilepsy and Behavior</i> , <b>2021</b> , 122, 108152	3.2	1	
287	Longitudinal Evaluation of the Stability of Hand Function in Rett Syndrome. <i>Journal of Pediatrics</i> , <b>2021</b> , 237, 244-249.e3	3.6	1	
286	Prevalence estimates of mental health problems in children and adolescents with intellectual disability: A systematic review and meta-analysis. <i>Australian and New Zealand Journal of Psychiatry</i> , <b>2020</b> , 54, 970-984	2.6	25	
285	Stillbirth risk prediction using machine learning for a large cohort of births from Western Australia, 1980-2015. <i>Scientific Reports</i> , <b>2020</b> , 10, 5354	4.9	13	
284	Cyclin-dependent-like kinase 5 is required for pain signaling in human sensory neurons and mouse models. <i>Science Translational Medicine</i> , <b>2020</b> , 12,	17.5	4	
283	Expanding the phenotype of the CDKL5 deficiency disorder: Are seizures mandatory?. <i>American Journal of Medical Genetics, Part A</i> , <b>2020</b> , 182, 1217-1222	2.5	4	
282	Systematic Review and Meta-analysis: Mental Health in Children With Neurogenetic Disorders Associated With Intellectual Disability. <i>Journal of the American Academy of Child and Adolescent</i> Psychiatry, <b>2020</b> , 59, 1036-1048	7.2	17	
281	The contributions of fetal growth restriction and gestational age to developmental outcomes at 12 months of age: A cohort study. <i>Early Human Development</i> , <b>2020</b> , 142, 104951	2.2	2	
<b>2</b> 80	Gastrostomy and quality of life in children with intellectual disability: a qualitative study. <i>Archives of Disease in Childhood</i> , <b>2020</b> , 105, 969-974	2.2	2	

279	A preliminary investigation of the effects of prenatal alcohol exposure on facial morphology in children with Autism Spectrum Disorder. <i>Alcohol</i> , <b>2020</b> , 86, 75-80	2.7	3
278	Implementing telehealth support to increase physical activity in girls and women with Rett syndromeActivRett: protocol for a waitlist randomised controlled trial. <i>BMJ Open</i> , <b>2020</b> , 10, e042446	3	2
277	Hospital admissions in children with developmental disabilities from ethnic minority backgrounds. <i>Developmental Medicine and Child Neurology</i> , <b>2020</b> , 62, 470-476	3.3	1
276	Pregnancy and birth outcomes of mothers with intellectual disability and their infants: Advocacy needed to improve well-being. <i>Disability and Health Journal</i> , <b>2020</b> , 13, 100871	4.2	4
275	Risk of Hospitalizations Following Gastrostomy in Children with Intellectual Disability. <i>Journal of Pediatrics</i> , <b>2020</b> , 217, 131-138.e10	3.6	3
274	Assessment of a Clinical Trial Metric for Rett Syndrome: Critical Analysis of the Rett Syndrome Behaviour Questionnaire. <i>Pediatric Neurology</i> , <b>2020</b> , 111, 4	2.9	3
273	Reliability of the Quality of Life Inventory-Disability Measure in Children with Intellectual Disability. Journal of Developmental and Behavioral Pediatrics, <b>2020</b> , 41, 534-539	2.4	7
272	Using directed-content analysis to identify a framework for understanding quality of life in adults with Rett syndrome. <i>Disability and Rehabilitation</i> , <b>2020</b> , 42, 3800-3807	2.4	3
271	Survival of children and adolescents with intellectual disability following gastrostomy insertion. Journal of Intellectual Disability Research, 2020, 64, 497-511	3.2	O
<b>27</b> 0	Are preterm birth and intra-uterine growth restriction more common in Western Australian children of immigrant backgrounds? A population based data linkage study. <i>BMC Pregnancy and Childbirth</i> , <b>2019</b> , 19, 287	3.2	3
269	Predicting Long-Term Survival Without Major Disability for Infants Born Preterm. <i>Journal of Pediatrics</i> , <b>2019</b> , 215, 90-97.e1	3.6	5
268	Very Early Identification and Intervention for Infants at Risk of Neurodevelopmental Disorders: AlTransdiagnostic Approach. <i>Child Development Perspectives</i> , <b>2019</b> , 13, 97-103	5.5	17
267	The Brain Basis of Comorbidity in Neurodevelopmental Disorders. <i>Current Developmental Disorders Reports</i> , <b>2019</b> , 6, 9-18	1.9	4
266	Content validation of the Quality of Life Inventory-Disability. <i>Child: Care, Health and Development</i> , <b>2019</b> , 45, 654-659	2.8	9
265	Genetic and epigenetic influences on the phenotype of Rett syndrome <b>2019</b> , 183-217		
264	Diagnosis of Autism Spectrum Disorder According to Maternal-Race Ethnicity and Country of Birth: A Register-Based Study. <i>Journal of Autism and Developmental Disorders</i> , <b>2019</b> , 49, 3611-3624	4.6	5
263	Birth seasonality and risk of autism spectrum disorder. European Journal of Epidemiology, 2019, 34, 785	7 <b>92</b> 1	13
262	Severity Assessment in CDKL5 Deficiency Disorder. <i>Pediatric Neurology</i> , <b>2019</b> , 97, 38-42	2.9	18

261	Recurrence Risk of Autism in Siblings and Cousins: AlMultinational, Population-Based Study. <i>Journal of the American Academy of Child and Adolescent Psychiatry</i> , <b>2019</b> , 58, 866-875	7.2	17
260	Cyclin-Dependent Kinase-Like 5 Deficiency Disorder: Clinical Review. <i>Pediatric Neurology</i> , <b>2019</b> , 97, 18-2	. <b>5</b> 2.9	60
259	The incidence, prevalence and clinical features of MECP2 duplication syndrome in Australian children. <i>Journal of Paediatrics and Child Health</i> , <b>2019</b> , 55, 1315-1322	1.3	11
258	Cannabis for refractory epilepsy in children: A review focusing on CDKL5 Deficiency Disorder. <i>Epilepsy Research</i> , <b>2019</b> , 151, 31-39	3	14
257	Association of Genetic and Environmental Factors With Autism in a 5-Country Cohort. <i>JAMA Psychiatry</i> , <b>2019</b> , 76, 1035-1043	14.5	151
256	Requirements for improving health and well-being of children with Prader-Willi syndrome and their families. <i>Journal of Paediatrics and Child Health</i> , <b>2019</b> , 55, 1029-1037	1.3	5
255	Maltreatment Risk Among Children With Disabilities <b>2019</b> , 117-126		
254	Association between interpregnancy interval and adverse birth outcomes in women with a previous stillbirth: an international cohort study. <i>Lancet, The</i> , <b>2019</b> , 393, 1527-1535	40	24
253	Assessing the quality, efficiency and usefulness of the Western Australian population-based Intellectual Disability Exploring Answers (IDEA) surveillance system: a surveillance system evaluation. <i>BMJ Open</i> , <b>2019</b> , 9, e026003	3	
252	Data Linkage: Canadian and Australian Perspectives on a Valuable Methodology for Intellectual and Developmental Disability Research. <i>Intellectual and Developmental Disabilities</i> , <b>2019</b> , 57, 439-462	2	13
251	A Population-Based Matched-Sibling Analysis Estimating the Associations Between First Interpregnancy Interval and Birth Outcomes. <i>American Journal of Epidemiology</i> , <b>2019</b> , 188, 9-16	3.8	15
250	Longitudinal effects of caregiving on parental well-being: the example of Rett syndrome, a severe neurological disorder. <i>European Child and Adolescent Psychiatry</i> , <b>2019</b> , 28, 505-520	5.5	15
249	Regan et al. Reply to "Sibling Comparison Design in Birth-Spacing Studies". <i>American Journal of Epidemiology</i> , <b>2019</b> , 188, 22-23	3.8	
248	A framework for understanding quality of life domains in individuals with the CDKL5 deficiency disorder. <i>American Journal of Medical Genetics, Part A</i> , <b>2019</b> , 179, 249-256	2.5	4
247	Psychometric properties of the Quality of Life Inventory-Disability (QI-Disability) measure. <i>Quality of Life Research</i> , <b>2019</b> , 28, 783-794	3.7	22
246	Epidemiology of gastrostomy insertion for children and adolescents with intellectual disability. European Journal of Pediatrics, <b>2019</b> , 178, 351-361	4.1	8
245	Risk of Developmental Disorders in Children of Immigrant Mothers: A Population-Based Data Linkage Evaluation. <i>Journal of Pediatrics</i> , <b>2019</b> , 204, 275-284.e3	3.6	8
244	Apgar score and risk of autism. <i>European Journal of Epidemiology</i> , <b>2019</b> , 34, 105-114	12.1	15

243	Parent-observed thematic data on quality of life in children with autism spectrum disorder. <i>Autism</i> , <b>2019</b> , 23, 71-80	6.6	15
242	Patterns of sedentary time and ambulatory physical activity in a Danish population of girls and women with Rett syndrome. <i>Disability and Rehabilitation</i> , <b>2019</b> , 41, 133-141	2.4	8
241	Use of health services in the last year of life and cause of death in people with intellectual disability: a retrospective matched cohort study. <i>BMJ Open</i> , <b>2018</b> , 8, e020268	3	19
240	Respiratory morbidity in Rett syndrome: an observational study. <i>Developmental Medicine and Child Neurology</i> , <b>2018</b> , 60, 951-957	3.3	13
239	Validation of intellectual disability coding through hospital morbidity records using an intellectual disability population-based database in Western Australia. <i>BMJ Open</i> , <b>2018</b> , 8, e019113	3	15
238	Parent-reported health-related quality of life of children with Down syndrome: a descriptive study. Developmental Medicine and Child Neurology, 2018, 60, 402-408	3.3	13
237	Sleep disturbances in Rett syndrome: Impact and management including use of sleep hygiene practices. <i>American Journal of Medical Genetics, Part A</i> , <b>2018</b> , 176, 1569-1577	2.5	19
236	Choice making in Rett syndrome: a descriptive study using video data. <i>Disability and Rehabilitation</i> , <b>2018</b> , 40, 813-819	2.4	8
235	Risk of stillbirth, preterm delivery, and fetal growth restriction following exposure in a previous birth: systematic review and meta-analysis. <i>BJOG: an International Journal of Obstetrics and Gynaecology</i> , <b>2018</b> , 125, 183-192	3.7	49
234	The prevalence of mental health disorders and symptoms in children and adolescents with cerebral palsy: a systematic review and meta-analysis. <i>Developmental Medicine and Child Neurology</i> , <b>2018</b> , 60, 30-38	3.3	48
233	What effect does regular exercise have on oxidative stress in people with Down syndrome? A systematic review with meta-analyses. <i>Journal of Science and Medicine in Sport</i> , <b>2018</b> , 21, 596-603	4.4	3
232	Vagus nerve stimulation for the treatment of refractory epilepsy in the CDKL5 Deficiency Disorder. <i>Epilepsy Research</i> , <b>2018</b> , 146, 36-40	3	22
231	Environmental enrichment intervention for Rett syndrome: an individually randomised stepped wedge trial. <i>Orphanet Journal of Rare Diseases</i> , <b>2018</b> , 13, 3	4.2	35
230	Evolving Trends of Gastrostomy Insertion Within a Pediatric Population. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2018</b> , 67, e89-e94	2.8	14
229	An investigation of the determinants of quality of life in adolescents and young adults with Down syndrome. <i>PLoS ONE</i> , <b>2018</b> , 13, e0197394	3.7	13
228	Risk and protective factors for the health of primary care-givers of children with autism spectrum disorders or intellectual disability: a narrative review. <i>Journal of Autism</i> , <b>2018</b> , 5, 1	0	5
227	Comparing Parental Well-Being and Its Determinants Across Three Different Genetic Disorders Causing Intellectual Disability. <i>Journal of Autism and Developmental Disorders</i> , <b>2018</b> , 48, 1651-1665	4.6	12
226	The Risk of Neurodevelopmental Disabilities in Children of Immigrant and Refugee Parents: Current Knowledge and Directions for Future Research. <i>Review Journal of Autism and Developmental Disorders</i> , <b>2018</b> , 5, 29-42	3.4	12

225	Intellectual Disability in Children Conceived Using Assisted Reproductive Technology. <i>Pediatrics</i> , <b>2018</b> , 142,	7.4	15
224	Oral health experiences of individuals with Rett syndrome: a retrospective study. <i>BMC Oral Health</i> , <b>2018</b> , 18, 195	3.7	5
223	Impact of Gastrostomy Placement on Nutritional Status, Physical Health, and Parental Well-Being of Females with Rett Syndrome: A Longitudinal Study of an Australian Population. <i>Journal of Pediatrics</i> , <b>2018</b> , 200, 188-195.e1	3.6	5
222	Expanding the clinical picture of the MECP2 Duplication syndrome. Clinical Genetics, 2017, 91, 557-563	4	32
221	Maltreatment Risk Among Children With Disabilities. <i>Pediatrics</i> , <b>2017</b> , 139,	7.4	62
220	Quantification of walking-based physical activity and sedentary time in individuals with Rett syndrome. <i>Developmental Medicine and Child Neurology</i> , <b>2017</b> , 59, 605-611	3.3	21
219	Use of the ketogenic diet to manage refractory epilepsy in CDKL5 disorder: Experience of >100 patients. <i>Epilepsia</i> , <b>2017</b> , 58, 1415-1422	6.4	30
218	Clinical and biological progress over 50 years in Rett syndrome. <i>Nature Reviews Neurology</i> , <b>2017</b> , 13, 37	-515	120
217	Risk of Mortality into Adulthood According to Gestational Age at Birth. <i>Journal of Pediatrics</i> , <b>2017</b> , 190, 185-191.e1	3.6	11
216	Feasibility[bf[Assessing[Diet[with[a[Mobile[Food[Record[for[Adolescents[and[Young[Adults[with[] Down[Syndrome. <i>Nutrients</i> , <b>2017</b> , 9,	6.7	6
215	Measurement of Sedentary Behaviors or "Downtime" in Rett Syndrome. <i>Journal of Child Neurology</i> , <b>2017</b> , 32, 1009-1013	2.5	6
214	Twenty-Five Year Survival of Children with Intellectual Disability in Western Australia. <i>Journal of Pediatrics</i> , <b>2017</b> , 188, 232-239.e2	3.6	13
213	Exploring quality of life of children with cerebral palsy and intellectual disability: What are the important domains of life?. <i>Child: Care, Health and Development</i> , <b>2017</b> , 43, 854-860	2.8	25
212	Ketogenic diet to manage refractory epilepsy in the cdkl5 disorder. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2017</b> , 88, e1.27-e1	5.5	
211	Autonomic breathing abnormalities in Rett syndrome: caregiver perspectives in an international database study. <i>Journal of Neurodevelopmental Disorders</i> , <b>2017</b> , 9, 15	4.6	20
210	Impacts of caring for a child with the CDKL5 disorder on parental wellbeing and family quality of life. <i>Orphanet Journal of Rare Diseases</i> , <b>2017</b> , 12, 16	4.2	29
209	Australian children living with rare diseases: experiences of diagnosis and perceived consequences of diagnostic delays. <i>Orphanet Journal of Rare Diseases</i> , <b>2017</b> , 12, 68	4.2	66
208	A qualitative investigation of recovery after femoral fracture in Rett syndrome. <i>Child: Care, Health and Development</i> , <b>2017</b> , 43, 232-239	2.8	3

207	Building the repertoire of measures of walking in Rett syndrome. <i>Disability and Rehabilitation</i> , <b>2017</b> , 39, 1926-1931	2.4	14
206	variants: Improving our understanding of a rare neurologic disorder. <i>Neurology: Genetics</i> , <b>2017</b> , 3, e200	3.8	27
205	Rare disease: a national survey of paediatriciansSexperiences and needs. <i>BMJ Paediatrics Open</i> , <b>2017</b> , 1, e000172	2.4	21
204	Qualitative Analysis of Parental Observations on Quality of Life in Australian Children with Down Syndrome. <i>Journal of Developmental and Behavioral Pediatrics</i> , <b>2017</b> , 38, 161-168	2.4	24
203	Caesarean section and risk of autism across gestational age: a multi-national cohort study of 5 million births. <i>International Journal of Epidemiology</i> , <b>2017</b> , 46, 429-439	7.8	31
202	Autism risk associated with parental age and with increasing difference in age between the parents. <i>Molecular Psychiatry</i> , <b>2016</b> , 21, 693-700	15.1	129
201	Parental perspectives on the communication abilities of their daughters with Rett syndrome. <i>Developmental Neurorehabilitation</i> , <b>2016</b> , 19, 17-25	1.8	19
200	Functional abilities in children and adults with the CDKL5 disorder. <i>American Journal of Medical Genetics, Part A</i> , <b>2016</b> , 170, 2860-2869	2.5	36
199	Seizure variables and their relationship to genotype and functional abilities in the CDKL5 disorder. <i>Neurology</i> , <b>2016</b> , 87, 2206-2213	6.5	51
198	Quantitative and qualitative insights into the experiences of children with Rett syndrome and their families. <i>Wiener Medizinische Wochenschrift</i> , <b>2016</b> , 166, 338-45	2.9	12
197	Twenty-five-year survival for aboriginal and caucasian children with congenital heart defects in Western Australia, 1980 to 2010. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , <b>2016</b> , 106, 1016-1031		4
196	Health care utilization and costs for children and adults with duchenne muscular dystrophy. <i>Muscle and Nerve</i> , <b>2016</b> , 53, 877-84	3.4	13
195	Prevalence and onset of comorbidities in the CDKL5 disorder differ from Rett syndrome. <i>Orphanet Journal of Rare Diseases</i> , <b>2016</b> , 11, 39	4.2	51
194	ViPAR: a software platform for the Virtual Pooling and Analysis of Research Data. <i>International Journal of Epidemiology</i> , <b>2016</b> , 45, 408-416	7.8	33
193	Conceptualizing a quality of life framework for girls with Rett syndrome using qualitative methods. <i>American Journal of Medical Genetics, Part A</i> , <b>2016</b> , 170, 645-53	2.5	36
192	Onset of maternal psychiatric disorders after the birth of a child with autism spectrum disorder: A retrospective cohort study. <i>Autism</i> , <b>2016</b> , 20, 37-44	6.6	11
191	Improved Survival in Down Syndrome over the Last 60 Years and the Impact of Perinatal Factors in Recent Decades. <i>Journal of Pediatrics</i> , <b>2016</b> , 169, 214-20.e1	3.6	38
190	The Natural History of Scoliosis in Females With Rett Syndrome. <i>Spine</i> , <b>2016</b> , 41, 856-63	3.3	40

### (2015-2016)

189	Maternal Psychiatric Disorder and the Risk of Autism Spectrum Disorder or Intellectual Disability in Subsequent Offspring. <i>Journal of Autism and Developmental Disorders</i> , <b>2016</b> , 46, 523-33	4.6	9	
188	Family satisfaction following spinal fusion in Rett syndrome. <i>Developmental Neurorehabilitation</i> , <b>2016</b> , 19, 31-7	1.8	6	
187	Brief Report: Burden of Care in Mothers of Children with Autism Spectrum Disorder or Intellectual Disability. <i>Journal of Autism and Developmental Disorders</i> , <b>2016</b> , 46, 1103-9	4.6	18	
186	Clinical Guidelines for Management of Bone Health in Rett Syndrome Based on Expert Consensus and Available Evidence. <i>PLoS ONE</i> , <b>2016</b> , 11, e0146824	3.7	33	
185	Young People with Intellectual Disability Transitioning to Adulthood: Do Behaviour Trajectories Differ in Those with and without Down Syndrome?. <i>PLoS ONE</i> , <b>2016</b> , 11, e0157667	3.7	44	
184	Validating the Rett Syndrome Gross Motor Scale. <i>PLoS ONE</i> , <b>2016</b> , 11, e0147555	3.7	42	
183	How can clinical ethics guide the management of comorbidities in the child with Rett syndrome?. Journal of Paediatrics and Child Health, <b>2016</b> , 52, 809-13	1.3	1	
182	Surgical fusion of early onset severe scoliosis increases survival in Rett syndrome: a cohort study. <i>Developmental Medicine and Child Neurology</i> , <b>2016</b> , 58, 632-8	3.3	19	
181	Population-Based Prevalence of Intellectual Disability and Autism Spectrum Disorders in Western Australia: A Comparison With Previous Estimates. <i>Medicine (United States)</i> , <b>2016</b> , 95, e3737	1.8	64	
180	Mothers of Children with Autism have Different Rates of Cancer According to the Presence of Intellectual Disability in Their Child. <i>Journal of Autism and Developmental Disorders</i> , <b>2016</b> , 46, 3106-14	4.6	3	
179	Determinants of sleep disturbances in Rett syndrome: Novel findings in relation to genotype. <i>American Journal of Medical Genetics, Part A</i> , <b>2016</b> , 170, 2292-300	2.5	23	
178	An Exploration of the Use of Eye Gaze and Gestures in Females With Rett Syndrome. <i>Journal of Speech, Language, and Hearing Research</i> , <b>2016</b> , 59, 1373-1383	2.8	10	
177	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. <i>Lancet Infectious Diseases, The</i> , <b>2016</b> , 16, 952-61	25.5	70	
176	Transition to adulthood for young people with intellectual disability: the experiences of their families. <i>European Child and Adolescent Psychiatry</i> , <b>2016</b> , 25, 1369-1381	5.5	39	
175	There is variability in the attainment of developmental milestones in the CDKL5 disorder. <i>Journal of Neurodevelopmental Disorders</i> , <b>2015</b> , 7, 2	4.6	47	
174	Longitudinal bone mineral content and density in Rett syndrome and their contributing factors. <i>Bone</i> , <b>2015</b> , 74, 191-8	4.7	11	
173	Rett syndrome: establishing a novel outcome measure for walking activity in an era of clinical trials for rare disorders. <i>Disability and Rehabilitation</i> , <b>2015</b> , 37, 1992-6	2.4	21	
172	The trajectories of sleep disturbances in Rett syndrome. <i>Journal of Sleep Research</i> , <b>2015</b> , 24, 223-33	5.8	59	

171	The increasing prevalence of reported diagnoses of childhood psychiatric disorders: a descriptive multinational comparison. <i>European Child and Adolescent Psychiatry</i> , <b>2015</b> , 24, 173-83	5.5	147
170	Patterns of depressive symptoms and social relating behaviors differ over time from other behavioral domains for young people with Down syndrome. <i>Medicine (United States)</i> , <b>2015</b> , 94, e710	1.8	14
169	A validation study of a modified Bouchard activity record that extends the concept of SuptimeSto Rett syndrome. <i>Developmental Medicine and Child Neurology</i> , <b>2015</b> , 57, 1137-42	3.3	9
168	Aspects of speech-language abilities are influenced by MECP2 mutation type in girls with Rett syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2015</b> , 167A, 354-62	2.5	24
167	Changes in Caesarean Delivery Rates in Western Australia from 1995 to 2010 by Gestational Age at Birth. <i>Paediatric and Perinatal Epidemiology</i> , <b>2015</b> , 29, 290-8	2.7	4
166	Spinal fusion in girls with Rett syndrome: post-operative recovery and family experiences. <i>Child: Care, Health and Development,</i> <b>2015</b> , 41, 1000-9	2.8	13
165	Maternal mental health and risk of child protection involvement: mental health diagnoses associated with increased risk. <i>Journal of Epidemiology and Community Health</i> , <b>2015</b> , 69, 1175-83	5.1	45
164	Onset of maternal psychiatric disorders after the birth of a child with intellectual disability: a retrospective cohort study. <i>Journal of Psychiatric Research</i> , <b>2015</b> , 61, 223-30	5.2	17
163	Association of Gestational Age at Birth with Reasons for Subsequent Hospitalisation: 18 Years of Follow-Up in a Western Australian Population Study. <i>PLoS ONE</i> , <b>2015</b> , 10, e0130535	3.7	27
162	Gastrointestinal dysmotility in Rett syndrome. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2014</b> , 58, 237-44	2.8	36
161	Experience of gastrostomy using a quality care framework: the example of rett syndrome. <i>Medicine</i> (United States), <b>2014</b> , 93, e328	1.8	19
160	Early mortality and primary causes of death in mothers of children with intellectual disability or autism spectrum disorder: a retrospective cohort study. <i>PLoS ONE</i> , <b>2014</b> , 9, e113430	3.7	14
159	Community participation for girls and women living with Rett syndrome. <i>Disability and Rehabilitation</i> , <b>2014</b> , 36, 894-9	2.4	12
158	Perspectives on hand function in girls and women with Rett syndrome. <i>Developmental Neurorehabilitation</i> , <b>2014</b> , 17, 210-7	1.8	14
157	Prevalence, clinical investigation, and management of gallbladder disease in Rett syndrome. <i>Developmental Medicine and Child Neurology</i> , <b>2014</b> , 56, 756-62	3.3	12
156	Community participation: Conversations with parent-carers of young women with Rett syndrome This manuscript was accepted under the Editorship of Susan Balandin. View all notes. <i>Journal of Intellectual and Developmental Disability</i> , <b>2014</b> , 39, 243-252	1.9	10
155	Twenty years of surveillance in Rett syndrome: what does this tell us?. <i>Orphanet Journal of Rare Diseases</i> , <b>2014</b> , 9, 87	4.2	72
154	Day occupation is associated with psychopathology for adolescents and young adults with Down syndrome. <i>BMC Psychiatry</i> , <b>2014</b> , 14, 266	4.2	8

#### (2013-2014)

153	"I have a good life": the meaning of well-being from the perspective of young adults with Down syndrome. <i>Disability and Rehabilitation</i> , <b>2014</b> , 36, 1290-8	2.4	31
152	Hospital admissions and gestational age at birth: 18 years of follow up in Western Australia. <i>Paediatric and Perinatal Epidemiology</i> , <b>2014</b> , 28, 536-44	2.7	19
151	Relationship between family quality of life and day occupations of young people with Down syndrome. <i>Social Psychiatry and Psychiatric Epidemiology</i> , <b>2014</b> , 49, 1455-65	4.5	20
150	Health conditions and their impact among adolescents and young adults with Down syndrome. <i>PLoS ONE</i> , <b>2014</b> , 9, e96868	3.7	52
149	Influence of the environment on participation in social roles for young adults with down syndrome. <i>PLoS ONE</i> , <b>2014</b> , 9, e108413	3.7	20
148	Dental admissions in children under two yearsa total-population investigation. <i>Child: Care, Health and Development</i> , <b>2013</b> , 39, 253-9	2.8	9
147	The International Collaboration for Autism Registry Epidemiology (iCARE): multinational registry-based investigations of autism risk factors and trends. <i>Journal of Autism and Developmental Disorders</i> , <b>2013</b> , 43, 2650-63	4.6	20
146	Neonatal outcomes after preterm birth by mothersShealth insurance status at birth: a retrospective cohort study. <i>BMC Health Services Research</i> , <b>2013</b> , 13, 40	2.9	7
145	Functioning and post-school transition outcomes for young people with Down syndrome. <i>Child:</i> Care, Health and Development, <b>2013</b> , 39, 789-800	2.8	25
144	The CDKL5 disorder is an independent clinical entity associated with early-onset encephalopathy. <i>European Journal of Human Genetics</i> , <b>2013</b> , 21, 266-73	5.3	161
143	Pubertal trajectory in females with Rett syndrome: a population-based study. <i>Brain and Development</i> , <b>2013</b> , 35, 912-20	2.2	8
142	Intellectual disability: population-based estimates of the proportion attributable to maternal alcohol use disorder during pregnancy. <i>Developmental Medicine and Child Neurology</i> , <b>2013</b> , 55, 271-7	3.3	41
141	Changes in risk factors for preterm birth in Western Australia 1984-2006. <i>BJOG: an International Journal of Obstetrics and Gynaecology</i> , <b>2013</b> , 120, 1051-60	3.7	23
140	Early development and regression in Rett syndrome. Clinical Genetics, 2013, 84, 572-6	4	29
139	Hospitalisation rates for children with intellectual disability or autism born in Western Australia 1983-1999: a population-based cohort study. <i>BMJ Open</i> , <b>2013</b> , 3,	3	27
138	Neonatal complications in public and private patients: a retrospective cohort study. <i>BMJ Open</i> , <b>2013</b> , 3,	3	8
137	Caring for a child with severe intellectual disability in China: the example of Rett syndrome. <i>Disability and Rehabilitation</i> , <b>2013</b> , 35, 343-51	2.4	10
136	Assessment and management of nutrition and growth in Rett syndrome. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2013</b> , 57, 451-60	2.8	37

135	AuthorsSreply: changes in risk factors for preterm birth in Western Australia 1984-2006. <i>BJOG: an International Journal of Obstetrics and Gynaecology</i> , <b>2013</b> , 120, 1699	3.7	
134	Resourceful and creative methods are necessary to research rare disorders. <i>Developmental Medicine and Child Neurology</i> , <b>2013</b> , 55, 870-1	3.3	3
133	Using a large international sample to investigate epilepsy in Rett syndrome. <i>Developmental Medicine and Child Neurology</i> , <b>2013</b> , 55, 553-8	3.3	47
132	The experiences of mothers of young adults with an intellectual disability transitioning from secondary school to adult life. <i>Journal of Intellectual and Developmental Disability</i> , <b>2013</b> , 38, 149-62	1.9	31
131	Role of public and private funding in the rising caesarean section rate: a cohort study. <i>BMJ Open</i> , <b>2013</b> , 3,	3	32
130	Trends in pre-existing mental health disorders among parents of infants born in Western Australia from 1990 to 2005. <i>Medical Journal of Australia</i> , <b>2013</b> , 198, 485-8	4	15
129	Hospital admissions in children with down syndrome: experience of a population-based cohort followed from birth. <i>PLoS ONE</i> , <b>2013</b> , 8, e70401	3.7	44
128	Application of Population-Based Linked Data to the Study of Intellectual Disability and Autism. <i>International Review of Research in Developmental Disabilities</i> , <b>2013</b> , 281-327	1	9
127	Maternal conditions and perinatal characteristics associated with autism spectrum disorder and intellectual disability. <i>PLoS ONE</i> , <b>2013</b> , 8, e50963	3.7	78
126	Family functioning in families with a child with Down syndrome: a mixed methods approach. <i>Journal of Intellectual Disability Research</i> , <b>2012</b> , 56, 961-73	3.2	55
125	Barriers to diagnosis of a rare neurological disorder in Chinalived experiences of Rett syndrome families. <i>American Journal of Medical Genetics, Part A</i> , <b>2012</b> , 158A, 1-9	2.5	13
124	Genetic drift. The diagnostic odyssey to Rett syndrome: the experience of an Australian family. <i>American Journal of Medical Genetics, Part A</i> , <b>2012</b> , 158A, 10-2	2.5	12
123	Initial assessment of the StepWatch Activity MonitorIto measure walking activity in Rett syndrome. <i>Disability and Rehabilitation</i> , <b>2012</b> , 34, 1010-5	2.4	14
122	"It's not what you were expecting, but it's still a beautiful journey": the experience of mothers of children with Down syndrome. <i>Disability and Rehabilitation</i> , <b>2012</b> , 34, 1501-10	2.4	35
121	What does the nature of the MECP2 mutation tell us about parental origin and recurrence risk in Rett syndrome?. <i>Clinical Genetics</i> , <b>2012</b> , 82, 526-33	4	7
120	Young adults with intellectual disability transitioning from school to post-school: a literature review framed within the ICF. <i>Disability and Rehabilitation</i> , <b>2012</b> , 34, 1747-64	2.4	75
119	The phenotype associated with a large deletion on MECP2. <i>European Journal of Human Genetics</i> , <b>2012</b> , 20, 921-7	5.3	25
118	To Feel Belonged: The Voices of Children and Youth with Disabilities on the Meaning of Wellbeing. <i>Child Indicators Research</i> , <b>2012</b> , 5, 375-391	1.9	54

## (2010-2011)

117	The relationship between MECP2 mutation type and health status and service use trajectories over time in a Rett syndrome population. <i>Research in Autism Spectrum Disorders</i> , <b>2011</b> , 5, 442-449	3	14
116	Use of equipment and respite services and caregiver health among Australian families living with Rett syndrome. <i>Research in Autism Spectrum Disorders</i> , <b>2011</b> , 5, 722-732	3	14
115	Measuring use and cost of health sector and related care in a population of girls and young women with Rett syndrome. <i>Research in Autism Spectrum Disorders</i> , <b>2011</b> , 5, 901-909	3	8
114	Total population investigation of dental hospitalizations in indigenous children under five years in Western Australia using linked data. <i>Australian Dental Journal</i> , <b>2011</b> , 56, 358-64	2.3	12
113	Autism and intellectual disability are differentially related to sociodemographic background at birth. <i>PLoS ONE</i> , <b>2011</b> , 6, e17875	3.7	82
112	P2-542 Population level investigation of hospital dental admissions for children under 5 years with intellectual disability. <i>Journal of Epidemiology and Community Health</i> , <b>2011</b> , 65, A371-A371	5.1	
111	Variation over time in medical conditions and health service utilization of children with Down syndrome. <i>Journal of Pediatrics</i> , <b>2011</b> , 158, 194-200.e1	3.6	43
110	Direct health care costs of children and adolescents with Down syndrome. <i>Journal of Pediatrics</i> , <b>2011</b> , 159, 541-5	3.6	26
109	A comparison of autism prevalence trends in Denmark and Western Australia. <i>Journal of Autism and Developmental Disorders</i> , <b>2011</b> , 41, 1601-8	4.6	51
108	Bone mineral content and density in Rett syndrome and their contributing factors. <i>Pediatric Research</i> , <b>2011</b> , 69, 293-8	3.2	36
107	Trends in the diagnosis of Rett syndrome in Australia. <i>Pediatric Research</i> , <b>2011</b> , 70, 313-9	3.2	90
106	Leisure participation for school-aged children with Down syndrome. <i>Disability and Rehabilitation</i> , <b>2011</b> , 33, 1880-9	2.4	46
105	Change in gross motor abilities of girls and women with rett syndrome over a 3- to 4-year period. Journal of Child Neurology, <b>2011</b> , 26, 1237-45	2.5	30
104	The impact of single gene and chromosomal disorders on hospital admissions of children and adolescents: a population-based study. <i>Public Health Genomics</i> , <b>2011</b> , 14, 153-61	1.9	20
103	Altered attainment of developmental milestones influences the age of diagnosis of rett syndrome. <i>Journal of Child Neurology</i> , <b>2011</b> , 26, 980-7	2.5	33
102	Longitudinal hand function in Rett syndrome. <i>Journal of Child Neurology</i> , <b>2011</b> , 26, 334-40	2.5	15
101	Evaluation of the processes of family-centred care for young children with intellectual disability in Western Australia. <i>Child: Care, Health and Development,</i> <b>2010</b> , 36, 709-18	2.8	19
100	Characteristics of non-Aboriginal and Aboriginal children and families with substantiated child maltreatment: a population-based study. <i>International Journal of Epidemiology</i> , <b>2010</b> , 39, 921-8	7.8	42

99	Epilepsy and mental retardation limited to females with PCDH19 mutations can present de novo or in single generation families. <i>Journal of Medical Genetics</i> , <b>2010</b> , 47, 211-6	5.8	64
98	Updating the profile of C-terminal MECP2 deletions in Rett syndrome. <i>Journal of Medical Genetics</i> , <b>2010</b> , 47, 242-8	5.8	51
97	Overview of Health Issues in School-aged Children with Down Syndrome. <i>International Review of Research in Mental Retardation</i> , <b>2010</b> , 39, 67-106		7
96	Valproate and risk of fracture in Rett syndrome. Archives of Disease in Childhood, <b>2010</b> , 95, 444-8	2.2	29
95	Rates and types of hospitalisations for children who have subsequent contact with the child protection system: a population based case-control study. <i>Journal of Epidemiology and Community Health</i> , <b>2010</b> , 64, 784-8	5.1	22
94	Unpacking the complex nature of the autism epidemic. <i>Research in Autism Spectrum Disorders</i> , <b>2010</b> , 4, 548-554	3	51
93	Monitoring child abuse and neglect at a population level: patterns of hospital admissions for maltreatment and assault. <i>Child Abuse and Neglect</i> , <b>2010</b> , 34, 823-32	4.3	27
92	Linking MECP2 and pain sensitivity: the example of Rett syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2010</b> , 152A, 1197-205	2.5	62
91	Atypical presentations and specific genotypes are associated with a delay in diagnosis in females with Rett syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2010</b> , 152A, 2535-42	2.5	27
90	Rett syndrome: revised diagnostic criteria and nomenclature. <i>Annals of Neurology</i> , <b>2010</b> , 68, 944-50	9.4	804
89	Stereotypical hand movements in 144 subjects with Rett syndrome from the population-based Australian database. <i>Movement Disorders</i> , <b>2010</b> , 25, 282-8	7	42
88	Hip displacement and scoliosis in Rett syndrome - screening is required. <i>Developmental Medicine and Child Neurology</i> , <b>2010</b> , 52, 93-8	3.3	10
87	Level of purposeful hand function as a marker of clinical severity in Rett syndrome. <i>Developmental Medicine and Child Neurology</i> , <b>2010</b> , 52, 817-23	3.3	37
86	Survival with Rett syndrome: comparing Rett's original sample with data from the Australian Rett Syndrome Database. <i>Developmental Medicine and Child Neurology</i> , <b>2010</b> , 52, 962-5	3.3	35
85	The common BDNF polymorphism may be a modifier of disease severity in Rett syndrome. <i>Neurology</i> , <b>2009</b> , 72, 1242-7	6.5	59
84	Parental experiences of scoliosis management in Rett syndrome. <i>Disability and Rehabilitation</i> , <b>2009</b> , 31, 1917-24	2.4	10
83	Autism spectrum disorders in young children: effect of changes in diagnostic practices. <i>International Journal of Epidemiology</i> , <b>2009</b> , 38, 1245-54	7.8	101
82	Factors associated with dental admissions for children aged under 5 years in Western Australia.  Archives of Disease in Childhood, <b>2009</b> , 94, 517-23	2.2	21

#### (2007-2009)

81	Increasing prevalence of neonatal withdrawal syndrome: population study of maternal factors and child protection involvement. <i>Pediatrics</i> , <b>2009</b> , 123, e614-21	7.4	81
80	Siblings of children with disabilities: challenges and opportunities. <i>Acta Paediatrica, International Journal of Paediatrics</i> , <b>2009</b> , 98, 23-4	3.1	26
79	InterRett, a model for international data collection in a rare genetic disorder. <i>Research in Autism Spectrum Disorders</i> , <b>2009</b> , 3, 639-639	3	39
78	Impact of scoliosis surgery on activities of daily living in females with Rett syndrome. <i>Journal of Pediatric Orthopaedics</i> , <b>2009</b> , 29, 369-74	2.4	14
77	Guidelines for management of scoliosis in Rett syndrome patients based on expert consensus and clinical evidence. <i>Spine</i> , <b>2009</b> , 34, E607-17	3.3	50
76	The impact of having a sibling with an intellectual disability: parental perspectives in two disorders. <i>Journal of Intellectual Disability Research</i> , <b>2008</b> , 52, 216-29	3.2	75
75	Physical and mental health in mothers of children with Down syndrome. <i>Journal of Pediatrics</i> , <b>2008</b> , 153, 320-6	3.6	73
74	Rare childhood diseases: how should we respond?. <i>Archives of Disease in Childhood</i> , <b>2008</b> , 93, 1071-4	2.2	85
73	Investigating genotype-phenotype relationships in Rett syndrome using an international data set. <i>Neurology</i> , <b>2008</b> , 70, 868-75	6.5	180
72	Intellectual disability co-occurring with schizophrenia and other psychiatric illness: population-based study. <i>British Journal of Psychiatry</i> , <b>2008</b> , 193, 364-72	5.4	226
71	A guide for the assessment and management of vitamin D status in people with intellectual disability (developed as an AADDM Working Party initiative). <i>Journal of Intellectual and Developmental Disability</i> , <b>2008</b> , 33, 184-8	1.9	2
70	Early determinants of fractures in Rett syndrome. <i>Pediatrics</i> , <b>2008</b> , 121, 540-6	7.4	60
69	Association of birth outcomes and maternal, school, and neighborhood characteristics with subsequent numeracy achievement. <i>American Journal of Epidemiology</i> , <b>2008</b> , 168, 21-9	3.8	28
68	Gross motor profile in rett syndrome as determined by video analysis. <i>Neuropediatrics</i> , <b>2008</b> , 39, 205-10	1.6	47
67	The diagnosis of autism in a female: could it be Rett syndrome?. <i>European Journal of Pediatrics</i> , <b>2008</b> , 167, 661-9	4.1	48
66	Relation between intrauterine growth and subsequent intellectual disability in a ten-year population cohort of children in Western Australia. <i>American Journal of Epidemiology</i> , <b>2008</b> , 167, 103-11	3.8	59
65	Delineation of large deletions of the MECP2 gene in Rett syndrome patients, including a familial case with a male proband. <i>European Journal of Human Genetics</i> , <b>2007</b> , 15, 1218-29	5.3	40
64	Co-occurrence of birth defects and intellectual disability. <i>Paediatric and Perinatal Epidemiology</i> , <b>2007</b> , 21, 65-75	2.7	34

63	Medical aspects of school-aged children with Down syndrome. <i>Developmental Medicine and Child Neurology</i> , <b>2007</b> , 41, 683-688	3.3	4
62	A population-based approach to the investigation of osteopenia in Rett syndrome. <i>Developmental Medicine and Child Neurology</i> , <b>2007</b> , 41, 323-328	3.3	2
61	Sleep problems in Rett syndrome. Brain and Development, 2007, 29, 609-16	2.2	91
60	Seizures in Rett syndrome: an overview from a one-year calendar study. <i>European Journal of Paediatric Neurology</i> , <b>2007</b> , 11, 310-7	3.8	68
59	Development of a video-based evaluation tool in Rett syndrome. <i>Journal of Autism and Developmental Disorders</i> , <b>2007</b> , 37, 1636-46	4.6	31
58	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. <i>Journal of Medical Genetics</i> , <b>2007</b> , 44, 148-52	5.8	72
57	Feeding experiences and growth status in a Rett syndrome population. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2007</b> , 45, 582-90	2.8	35
56	NTNG1 mutations are a rare cause of Rett syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2006</b> , 140, 691-4	2.5	37
55	The association between behavior and genotype in Rett syndrome using the Australian Rett Syndrome Database. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , <b>2006</b> , 141B, 177-83	3.5	45
54	Predictors of scoliosis in Rett syndrome. <i>Journal of Child Neurology</i> , <b>2006</b> , 21, 809-13	2.5	51
53	Physical and mental health of mothers caring for a child with Rett syndrome. <i>Pediatrics</i> , <b>2006</b> , 118, e115	5 <del>2-</del> 64	57
52	Lost in translation: translational interference from a recurrent mutation in exon 1 of MECP2. Journal of Medical Genetics, <b>2006</b> , 43, 470-7	5.8	23
51	Early progressive encephalopathy in boys and MECP2 mutations. <i>Neurology</i> , <b>2006</b> , 67, 164-6	6.5	75
50	Maternal health in pregnancy and intellectual disability in the offspring: a population-based study. <i>Annals of Epidemiology</i> , <b>2006</b> , 16, 448-54	6.4	54
49	Rett syndrome in Australia: a review of the epidemiology. <i>Journal of Pediatrics</i> , <b>2006</b> , 148, 347-52	3.6	241
48	Predictors of seizure onset in Rett syndrome. <i>Journal of Pediatrics</i> , <b>2006</b> , 149, 542-7	3.6	79
47	Effectiveness of posthumous molecular diagnosis from a kept baby tooth. <i>Lancet, The</i> , <b>2005</b> , 366, 1584	40	3
46	InterRettThe application of bioinformatics to International Rett syndrome research. <i>Annals of Human Biology</i> , <b>2005</b> , 32, 228-36	1.7	11

#### (2002-2005)

45	IDEA (Intellectual Disability Exploring Answers): a population-based database for intellectual disability in Western Australia. <i>Annals of Human Biology</i> , <b>2005</b> , 32, 237-43	1.7	59
44	p.R270X MECP2 mutation and mortality in Rett syndrome. <i>European Journal of Human Genetics</i> , <b>2005</b> , 13, 1235-8	5.3	28
43	Genotype and early development in Rett syndrome: the value of international data. <i>Brain and Development</i> , <b>2005</b> , 27 Suppl 1, S59-S68	2.2	30
42	Association of sociodemographic characteristics of children with intellectual disability in Western Australia. <i>Social Science and Medicine</i> , <b>2005</b> , 60, 1499-513	5.1	79
41	Health service use in Rett syndrome. <i>Journal of Child Neurology</i> , <b>2005</b> , 20, 42-50	2.5	9
40	Hospitalisations from birth to 5 years in a population cohort of Western Australian children with intellectual disability. <i>Archives of Disease in Childhood</i> , <b>2005</b> , 90, 1243-8	2.2	27
39	Refining the phenotype of common mutations in Rett syndrome. <i>Journal of Medical Genetics</i> , <b>2004</b> , 41, 25-30	5.8	65
38	Trisomy 21 and Rett syndrome: a double burden. <i>Journal of Paediatrics and Child Health</i> , <b>2004</b> , 40, 406-9	9 1.3	6
37	Mutations of CDKL5 cause a severe neurodevelopmental disorder with infantile spasms and mental retardation. <i>American Journal of Human Genetics</i> , <b>2004</b> , 75, 1079-93	11	352
36	How can the Internet help parents of children with rare neurologic disorders?. <i>Journal of Child Neurology</i> , <b>2004</b> , 19, 902-7	2.5	31
35	Effects of MECP2 mutation type, location and X-inactivation in modulating Rett syndrome phenotype. <i>American Journal of Medical Genetics Part A</i> , <b>2003</b> , 118A, 103-14		84
34	Prevalence of intellectual disability in Western Australia. <i>Paediatric and Perinatal Epidemiology</i> , <b>2003</b> , 17, 58-67	2.7	76
33	Mutation analysis in the MECP2 gene and genetic counselling for Rett syndrome. <i>Journal of Medical Genetics</i> , <b>2003</b> , 40, 380-4	5.8	12
32	Describing the phenotype in Rett syndrome using a population database. <i>Archives of Disease in Childhood</i> , <b>2003</b> , 88, 38-43	2.2	88
31	Hand preference, extent of laterality, and functional hand use in Rett syndrome. <i>Journal of Child Neurology</i> , <b>2003</b> , 18, 481-7	2.5	13
30	InterRett and RettBASE: International Rett Syndrome Association databases for Rett syndrome. <i>Journal of Child Neurology</i> , <b>2003</b> , 18, 709-13	2.5	30
29	Patients with the R133C mutation: is their phenotype different from patients with Rett syndrome with other mutations?. <i>Journal of Medical Genetics</i> , <b>2003</b> , 40, e52	5.8	67
28	The epidemiology of mental retardation: challenges and opportunities in the new millennium.  Mental Retardation and Developmental Disabilities Research Reviews, 2002, 8, 117-34		428

27	Functional status of school-aged children with Down syndrome. <i>Journal of Paediatrics and Child Health</i> , <b>2002</b> , 38, 160-5	1.3	45
26	A case of multiple congenital anomalies in association with Rett syndrome confirmed by MECP2 mutation screening. <i>Clinical Dysmorphology</i> , <b>2001</b> , 10, 185-8	0.9	6
25	Using the Internet to pilot a questionnaire on childhood disability in Rett syndrome. <i>Child: Care, Health and Development</i> , <b>2001</b> , 27, 535-43	2.8	16
24	FriedreichS ataxia presenting after cardiac transplantation. <i>Archives of Disease in Childhood</i> , <b>2001</b> , 84, 167-8	2.2	17
23	Occurrence of Rett syndrome in boys. <i>Journal of Child Neurology</i> , <b>2001</b> , 16, 333-8	2.5	62
22	Guidelines for reporting clinical features in cases with MECP2 mutations. <i>Brain and Development</i> , <b>2001</b> , 23, 208-11	2.2	121
21	Medium-term open label trial of L-carnitine in Rett syndrome. <i>Brain and Development</i> , <b>2001</b> , 23 Suppl 1, S85-9	2.2	31
20	Sleep dysfunction in Rett syndrome: lack of age related decrease in sleep duration. <i>Brain and Development</i> , <b>2001</b> , 23 Suppl 1, S101-3	2.2	33
19	Functional status, medical impairments, and rehabilitation resources in 84 females with Rett syndrome: a snapshot across the world from the parental perspective. <i>Disability and Rehabilitation</i> , <b>2001</b> , 23, 107-17	2.4	44
18	Intellectual disability in Western Australia. <i>Journal of Paediatrics and Child Health</i> , <b>2000</b> , 36, 213-5	1.3	23
17	Family data in Rett syndrome: association with other genetic disorders. <i>Journal of Paediatrics and Child Health</i> , <b>2000</b> , 36, 336-9	1.3	2
16	Using genetic epidemiology to study Rett syndrome: the design of a case-control study. <i>Paediatric and Perinatal Epidemiology</i> , <b>2000</b> , 14, 85-95	2.7	5
15	Survival of infants born with Down's syndrome: 1980-96. <i>Paediatric and Perinatal Epidemiology</i> , <b>2000</b> , 14, 163-71	2.7	91
14	Long-read sequence analysis of the MECP2 gene in Rett syndrome patients: correlation of disease severity with mutation type and location. <i>Human Molecular Genetics</i> , <b>2000</b> , 9, 1119-29	5.6	196
13	Patterns of pregnancy loss, perinatal mortality, and postneonatal childhood deaths in families of girls with Rett syndrome. <i>Journal of Child Neurology</i> , <b>1999</b> , 14, 440-5	2.5	11
12	Rett syndrome: randomized controlled trial of L-carnitine. <i>Journal of Child Neurology</i> , <b>1999</b> , 14, 162-7	2.5	41
11	Prolonged QT interval in Rett syndrome. Archives of Disease in Childhood, 1999, 80, 470-2	2.2	49
10	Familial aggregation in Rett syndrome: what is the evidence for clustering of other disorders in families of affected girls?. <i>American Journal of Medical Genetics Part A</i> , <b>1999</b> , 82, 228-34		4

#### LIST OF PUBLICATIONS

9	Metacarpophalangeal pattern profile and bone age in Rett syndrome: further radiological clues to the diagnosis. <i>American Journal of Medical Genetics Part A</i> , <b>1999</b> , 83, 88-95		18	
8	A population-based approach to the investigation of osteopenia in Rett syndrome. <i>Developmental Medicine and Child Neurology</i> , <b>1999</b> , 41, 323-8	3.3	38	
7	Medical aspects of school-aged children with Down syndrome. <i>Developmental Medicine and Child Neurology</i> , <b>1999</b> , 41, 683-8	3.3	24	
6	Diagnosis of Rett syndrome: can a radiograph help?. <i>Developmental Medicine and Child Neurology</i> , <b>1998</b> , 40, 737-42	3.3	8	
5	Rett syndrome: significant clinical overlap with Angelman syndrome but not with methylation status. <i>Journal of Child Neurology</i> , <b>1998</b> , 13, 448-51	2.5	14	
4	Is the girl with Rett syndrome normal at birth?. <i>Developmental Medicine and Child Neurology</i> , <b>1998</b> , 40, 115-21	3.3	73	
3	The prevalence and incidence of Rett syndrome in Australia. <i>European Child and Adolescent Psychiatry</i> , <b>1997</b> , 6 Suppl 1, 8-10	5.5	33	
2	Skeletal abnormalities in Rett syndrome: increasing evidence for dysmorphogenetic defects. <i>American Journal of Medical Genetics Part A</i> , <b>1995</b> , 58, 282-5		19	
1	RettS syndrome: opportunity for Australian study. <i>Medical Journal of Australia</i> , <b>1993</b> , 159, 832	4	4	