Jennepher Anne Downs

List of Publications by Year in Descending Order

163 papers	2,737 citations	30 h-index	43 g-index
173 ext. papers	3,512 ext. citations	3.3 avg, IF	5.31 L-index

#	Paper	IF	Citations
163	Modifiable child and caregiver factors that influence community participation among children with Down syndrome.. *Disability and Rehabilitation*, **2022**, 44, 600-607	2.4	2
162	Family-Centered Telehealth Supporting Motor Skills and Activity in Individuals With Rett Syndrome. *Advances in Medical Technologies and Clinical Practice Book Series*, **2022**, 147-171	0.3	
161	The Lived Experience of ParentsRReceiving the Diagnosis of CDKL5 Deficiency Disorder for Their Child.. *Journal of Child Neurology*, **2022**, 8830738221076285	2.5	
160	Daytime sleepiness and emotional and behavioral disturbances in Prader-Willi syndrome.. *European Journal of Pediatrics*, **2022**, 1	4.1	
159	Improving clinical trial readiness to accelerate development of new therapeutics for Rett syndrome.. *Orphanet Journal of Rare Diseases*, **2022**, 17, 108	4.2	0
158	A brief history of MECP2 duplication syndrome: 20-years of clinical understanding.. *Orphanet Journal of Rare Diseases*, **2022**, 17, 131	4.2	0
157	Initial Validation and Reliability of the CDKL5 Deficiency Disorder Hand Function Scale (CDD-Hand).. *Journal of Child Neurology*, **2022**, 8830738221091044	2.5	0
156	Negative impact of insomnia and daytime sleepiness on quality of life in individuals with the cyclin-dependent kinase-like 5 deficiency disorder.. *Journal of Sleep Research*, **2022**, e13600	5.8	0
155	Medical Comorbidities in MECP2 Duplication Syndrome: Results from the International MECP2 Duplication Database. *Children*, **2022**, 9, 633	2.8	1
154	Parent and therapist perspectives on "uptime" activities and participation in Rett syndrome. *Disability and Rehabilitation*, **2021**, 1-8	2.4	
153	Determinants of quality of life in Rett syndrome: new findings on associations with genotype. *Journal of Medical Genetics*, **2021**, 58, 637-644	5.8	4
152	Using a trauma informed practice framework to enhance understanding of and identify support strategies for behavioural difficulties in young people with Prader-Willi syndrome. *Research in Developmental Disabilities*, **2021**, 110, 103839	2.7	1
151	Oral health care and service utilisation in individuals with Rett syndrome: an international cross-sectional study. *Journal of Intellectual Disability Research*, **2021**, 65, 561-576	3.2	2
150	A Pilot Study Delivering Physiotherapy Support for Rett Syndrome Using a Telehealth Framework Suitable for COVID-19 Lockdown. *Developmental Neurorehabilitation*, **2021**, 24, 429-434	1.8	3
149	Parent Carer Quality of Life and Night-Time Attendance in Non-Ambulant Youth with Neuromuscular Disorders. *Developmental Neurorehabilitation*, **2021**, 24, 456-465	1.8	1
148	Comorbidities and quality of life in children with intellectual disability. *Child: Care, Health and Development*, **2021**, 47, 654-666	2.8	1
147	Determinants of sleep problems in children with intellectual disability. *Journal of Sleep Research*, **2021**, 30, e13361	5.8	2

146	The effect of functioning on Quality of Life Inventory-Disability measured quality of life is not mediated or moderated by parental psychological distress. *Quality of Life Research*, **2021**, 30, 2875-2885	3.7	0
145	Caregiver-mediated interventions to support self-regulation among infants and young children (0-5 years): a protocol for a realist review. *BMJ Open*, **2021**, 11, e046078	3	2
144	Exploring genotype-phenotype relationships in the CDKL5 deficiency disorder using an international dataset. *Clinical Genetics*, **2021**, 99, 157-165	4	5
143	Exploring quality of life in individuals with a severe developmental and epileptic encephalopathy, CDKL5 Deficiency Disorder. *Epilepsy Research*, **2021**, 169, 106521	3	7
142	Functioning, participation, and quality of life in children with intellectual disability: an observational study. *Developmental Medicine and Child Neurology*, **2021**, 63, 89-96	3.3	12
141	Women Diagnosed with Ovarian Cancer: Patient and Carer Experiences and Perspectives. *Patient Related Outcome Measures*, **2021**, 12, 33-43	2.9	3
140	Decline in gross motor skills in adult Rett syndrome; results from a Danish longitudinal study. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3683-3693	2.5	0
139	Sleep-disordered breathing in Australian children with Prader-Willi syndrome following initiation of growth hormone therapy. *Journal of Paediatrics and Child Health*, **2021**,	1.3	1
138	Content Validation of Clinician-Reported Items for a Severity Measure for CDKL5 Deficiency Disorder. *Journal of Child Neurology*, **2021**, 36, 998-1006	2.5	2
137	The Rett Syndrome Gross Motor Scale - Dutch Version (RSGMS-NL) Can Reliably Assess Gross Motor Skills in Dutch Individuals with Rett Syndrome. *Developmental Neurorehabilitation*, **2021**, 1-7	1.8	0
136	Associations Between Hyperphagia, Symptoms of Sleep Breathing Disorder, Behaviour Difficulties and Caregiver Well-Being in Prader-Willi Syndrome: A Preliminary Study. *Journal of Autism and Developmental Disorders*, **2021**, 1	4.6	
135	The perceived effects of cannabis products in the management of seizures in CDKL5 Deficiency Disorder. *Epilepsy and Behavior*, **2021**, 122, 108152	3.2	1
134	Longitudinal Evaluation of the Stability of Hand Function in Rett Syndrome. *Journal of Pediatrics*, **2021**, 237, 244-249.e3	3.6	1
133	Early Motor Function of Children With Autism Spectrum Disorder: A Systematic Review. *Pediatrics*, **2021**, 147,	7.4	2
132	Feasibility and Effectiveness of an Individualized 12-Week "Uptime" Participation (U-PART) Intervention in Girls and Women With Rett Syndrome. *Physical Therapy*, **2020**, 100, 168-179	3.3	4
131	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
130	Cyclin-dependent-like kinase 5 is required for pain signaling in human sensory neurons and mouse models. *Science Translational Medicine*, **2020**, 12,	17.5	4
129	Benefits of powered standing wheelchair devices for adolescents with Duchenne muscular dystrophy in the first year of use. *Journal of Paediatrics and Child Health*, **2020**, 56, 1419-1425	1.3	6

128	Expanding the phenotype of the CDKL5 deficiency disorder: Are seizures mandatory?. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1217-1222	2.5	4
127	Exploring enablers and barriers to accessing health services after a fall among people with intellectual disability. *Journal of Applied Research in Intellectual Disabilities*, **2020**, 33, 604-617	2.2	1
126	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
125	The contributions of fetal growth restriction and gestational age to developmental outcomes at 12 months of age: A cohort study. *Early Human Development*, **2020**, 142, 104951	2.2	2
124	Gastrostomy and quality of life in children with intellectual disability: a qualitative study. *Archives of Disease in Childhood*, **2020**, 105, 969-974	2.2	2
123	A preliminary investigation of the effects of prenatal alcohol exposure on facial morphology in children with Autism Spectrum Disorder. *Alcohol*, **2020**, 86, 75-80	2.7	3
122	Comparing Web-Based Mindfulness With Loving-Kindness and Compassion Training for Promoting Well-Being in Pregnancy: Protocol for a Three-Arm Pilot Randomized Controlled Trial. *JMIR Research Protocols*, **2020**, 9, e19803	2	2
121	Implementing telehealth support to increase physical activity in girls and women with Rett syndromeActivRett: protocol for a waitlist randomised controlled trial. *BMJ Open*, **2020**, 10, e042446	3	2
120	Hospital admissions in children with developmental disabilities from ethnic minority backgrounds. *Developmental Medicine and Child Neurology*, **2020**, 62, 470-476	3.3	1
119	Risk of Hospitalizations Following Gastrostomy in Children with Intellectual Disability. *Journal of Pediatrics*, **2020**, 217, 131-138.e10	3.6	3
118	Assessment of a Clinical Trial Metric for Rett Syndrome: Critical Analysis of the Rett Syndrome Behaviour Questionnaire. *Pediatric Neurology*, **2020**, 111, 4	2.9	3
117	The development of a consensus statement for the prescription of powered wheelchair standing devices in Duchenne muscular dystrophy. *Disability and Rehabilitation*, **2020**, 1-9	2.4	4
116	Reliability of the Quality of Life Inventory-Disability Measure in Children with Intellectual Disability. *Journal of Developmental and Behavioral Pediatrics*, **2020**, 41, 534-539	2.4	7
115	Using directed-content analysis to identify a framework for understanding quality of life in adults with Rett syndrome. *Disability and Rehabilitation*, **2020**, 42, 3800-3807	2.4	3
114	Survival of children and adolescents with intellectual disability following gastrostomy insertion. *Journal of Intellectual Disability Research*, **2020**, 64, 497-511	3.2	0
113	Are preterm birth and intra-uterine growth restriction more common in Western Australian children of immigrant backgrounds? A population based data linkage study. *BMC Pregnancy and Childbirth*, **2019**, 19, 287	3.2	3
112	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
111	The Brain Basis of Comorbidity in Neurodevelopmental Disorders. *Current Developmental Disorders Reports*, **2019**, 6, 9-18	1.9	4

110	Content validation of the Quality of Life Inventory-Disability. *Child: Care, Health and Development*, **2019**, 45, 654-659	2.8	9
109	Genetic and epigenetic influences on the phenotype of Rett syndrome **2019**, 183-217		
108	Diagnosis of Autism Spectrum Disorder According to Maternal-Race Ethnicity and Country of Birth: A Register-Based Study. *Journal of Autism and Developmental Disorders*, **2019**, 49, 3611-3624	4.6	5
107	Severity Assessment in CDKL5 Deficiency Disorder. *Pediatric Neurology*, **2019**, 97, 38-42	2.9	18
106	Powered standing wheelchairs promote independence, health and community involvement in adolescents with Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2019**, 29, 221-230	2.9	10
105	The incidence, prevalence and clinical features of MECP2 duplication syndrome in Australian children. *Journal of Paediatrics and Child Health*, **2019**, 55, 1315-1322	1.3	11
104	Cannabis for refractory epilepsy in children: A review focusing on CDKL5 Deficiency Disorder. *Epilepsy Research*, **2019**, 151, 31-39	3	14
103	Requirements for improving health and well-being of children with Prader-Willi syndrome and their families. *Journal of Paediatrics and Child Health*, **2019**, 55, 1029-1037	1.3	5
102	Incidence and associated risk factors for falls in adults with intellectual disability. *Journal of Intellectual Disability Research*, **2019**, 63, 1441-1452	3.2	3
101	Data Linkage: Canadian and Australian Perspectives on a Valuable Methodology for Intellectual and Developmental Disability Research. *Intellectual and Developmental Disabilities*, **2019**, 57, 439-462	2	13
100	Incidence and prevalence of falls in adults with intellectual disability living in the community: a systematic review. *JBI Database of Systematic Reviews and Implementation Reports*, **2019**, 17, 390-413	1.6	10
99	Mental wellbeing in non-ambulant youth with neuromuscular disorders: What makes the difference?. *Neuromuscular Disorders*, **2019**, 29, 48-58	2.9	5
98	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
97	A framework for understanding quality of life domains in individuals with the CDKL5 deficiency disorder. *American Journal of Medical Genetics, Part A*, **2019**, 179, 249-256	2.5	4
96	Psychometric properties of the Quality of Life Inventory-Disability (QI-Disability) measure. *Quality of Life Research*, **2019**, 28, 783-794	3.7	22
95	Facilitators and Barriers of Participation in "Uptime" Activities in Girls and Women With Rett Syndrome: Perspectives From Parents and Professionals. *Qualitative Health Research*, **2019**, 29, 609-619	3.9	4
94	Epidemiology of gastrostomy insertion for children and adolescents with intellectual disability. *European Journal of Pediatrics*, **2019**, 178, 351-361	4.1	8
93	Risk of Developmental Disorders in Children of Immigrant Mothers: A Population-Based Data Linkage Evaluation. *Journal of Pediatrics*, **2019**, 204, 275-284.e3	3.6	8

92	Parent-observed thematic data on quality of life in children with autism spectrum disorder. *Autism*, **2019**, 23, 71-80	6.6	15
91	Patterns of sedentary time and ambulatory physical activity in a Danish population of girls and women with Rett syndrome. *Disability and Rehabilitation*, **2019**, 41, 133-141	2.4	8
90	Addressing challenges in gaining informed consent for a research study investigating falls in people with intellectual disability. *British Journal of Learning Disabilities*, **2018**, 46, 92-100	1	13
89	Respiratory morbidity in Rett syndrome: an observational study. *Developmental Medicine and Child Neurology*, **2018**, 60, 951-957	3.3	13
88	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
87	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
86	Choice making in Rett syndrome: a descriptive study using video data. *Disability and Rehabilitation*, **2018**, 40, 813-819	2.4	8
85	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
84	What effect does regular exercise have on oxidative stress in people with Down syndrome? A systematic review with meta-analyses. *Journal of Science and Medicine in Sport*, **2018**, 21, 596-603	4.4	3
83	Investigating falls in adults with intellectual disability living in community settings and their experiences of post-fall care services: protocol for a prospective observational cohort study. *BMC Geriatrics*, **2018**, 18, 171	4.1	3
82	Vagus nerve stimulation for the treatment of refractory epilepsy in the CDKL5 Deficiency Disorder. *Epilepsy Research*, **2018**, 146, 36-40	3	22
81	Environmental enrichment intervention for Rett syndrome: an individually randomised stepped wedge trial. *Orphanet Journal of Rare Diseases*, **2018**, 13, 3	4.2	35
80	Evolving Trends of Gastrostomy Insertion Within a Pediatric Population. *Journal of Pediatric Gastroenterology and Nutrition*, **2018**, 67, e89-e94	2.8	14
79	Comparing Parental Well-Being and Its Determinants Across Three Different Genetic Disorders Causing Intellectual Disability. *Journal of Autism and Developmental Disorders*, **2018**, 48, 1651-1665	4.6	12
78	The Risk of Neurodevelopmental Disabilities in Children of Immigrant and Refugee Parents: Current Knowledge and Directions for Future Research. *Review Journal of Autism and Developmental Disorders*, **2018**, 5, 29-42	3.4	12
77	Impact of biobanks on research outcomes in rare diseases: a systematic review. *Orphanet Journal of Rare Diseases*, **2018**, 13, 202	4.2	16
76	Oral health experiences of individuals with Rett syndrome: a retrospective study. *BMC Oral Health*, **2018**, 18, 195	3.7	5
75	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. *Journal of Pediatrics*, **2018**, 200, 188-195.e1	3.6	5

74	Expanding the clinical picture of the MECP2 Duplication syndrome. *Clinical Genetics*, **2017**, 91, 557-563	4	32
73	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
72	Use of the ketogenic diet to manage refractory epilepsy in CDKL5 disorder: Experience of >100 patients. *Epilepsia*, **2017**, 58, 1415-1422	6.4	30
71	Clinical and biological progress over 50 years in Rett syndrome. *Nature Reviews Neurology*, **2017**, 13, 37-51	15	120
70	Incidence and prevalence of falls in adults with intellectual disability living in the community: a systematic review protocol. *JBI Database of Systematic Reviews and Implementation Reports*, **2017**, 15, 1819-1823	1.6	3
69	Measurement of Sedentary Behaviors or "Downtime" in Rett Syndrome. *Journal of Child Neurology*, **2017**, 32, 1009-1013	2.5	6
68	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
67	Ketogenic diet to manage refractory epilepsy in the cdkl5 disorder. *Journal of Neurology, Neurosurgery and Psychiatry*, **2017**, 88, e1.27-e1	5.5	
66	Autonomic breathing abnormalities in Rett syndrome: caregiver perspectives in an international database study. *Journal of Neurodevelopmental Disorders*, **2017**, 9, 15	4.6	20
65	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
64	A qualitative investigation of recovery after femoral fracture in Rett syndrome. *Child: Care, Health and Development*, **2017**, 43, 232-239	2.8	3
63	Quality of Life and Psychosocial Well-Being in Youth With Neuromuscular Disorders Who Are Wheelchair Users: A Systematic Review. *Archives of Physical Medicine and Rehabilitation*, **2017**, 98, 1004-1017.e1	2.8	17
62	Building the repertoire of measures of walking in Rett syndrome. *Disability and Rehabilitation*, **2017**, 39, 1926-1931	2.4	14
61	variants: Improving our understanding of a rare neurologic disorder. *Neurology: Genetics*, **2017**, 3, e200	3.8	27
60	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
59	Parental perspectives on the communication abilities of their daughters with Rett syndrome. *Developmental Neurorehabilitation*, **2016**, 19, 17-25	1.8	19
58	Functional abilities in children and adults with the CDKL5 disorder. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2860-2869	2.5	36
57	Seizure variables and their relationship to genotype and functional abilities in the CDKL5 disorder. *Neurology*, **2016**, 87, 2206-2213	6.5	51

56	Quantitative and qualitative insights into the experiences of children with Rett syndrome and their families. *Wiener Medizinische Wochenschrift*, **2016**, 166, 338-45	2.9	12
55	Propulsion strategy in the gait of primary school children; the effect of age and speed. *Human Movement Science*, **2016**, 50, 54-61	2.4	5
54	Prevalence and onset of comorbidities in the CDKL5 disorder differ from Rett syndrome. *Orphanet Journal of Rare Diseases*, **2016**, 11, 39	4.2	51
53	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
52	The Natural History of Scoliosis in Females With Rett Syndrome. *Spine*, **2016**, 41, 856-63	3.3	40
51	Family satisfaction following spinal fusion in Rett syndrome. *Developmental Neurorehabilitation*, **2016**, 19, 31-7	1.8	6
50	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
49	Validating the Rett Syndrome Gross Motor Scale. *PLoS ONE*, **2016**, 11, e0147555	3.7	42
48	How can clinical ethics guide the management of comorbidities in the child with Rett syndrome?. *Journal of Paediatrics and Child Health*, **2016**, 52, 809-13	1.3	1
47	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
46	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
45	A fine balance and a shared learning journey: Exploring healthcare engagement through the experiences of youth with Neuromuscular Disorders. *NeuroRehabilitation*, **2016**, 39, 519-534	2	2
44	An Exploration of the Use of Eye Gaze and Gestures in Females With Rett Syndrome. *Journal of Speech, Language, and Hearing Research*, **2016**, 59, 1373-1383	2.8	10
43	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
42	There is variability in the attainment of developmental milestones in the CDKL5 disorder. *Journal of Neurodevelopmental Disorders*, **2015**, 7, 2	4.6	47
41	Longitudinal bone mineral content and density in Rett syndrome and their contributing factors. *Bone*, **2015**, 74, 191-8	4.7	11
40	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
39	The trajectories of sleep disturbances in Rett syndrome. *Journal of Sleep Research*, **2015**, 24, 223-33	5.8	59

38	A validation study of a modified Bouchard activity record that extends the concept of Ruptime to Rett syndrome. *Developmental Medicine and Child Neurology*, **2015**, 57, 1137-42	3.3	9
37	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
36	Spinal fusion in girls with Rett syndrome: post-operative recovery and family experiences. *Child: Care, Health and Development*, **2015**, 41, 1000-9	2.8	13
35	"The problem with running"--comparing the propulsion strategy of children with developmental coordination disorder and typically developing children. *Gait and Posture*, **2014**, 39, 547-52	2.6	16
34	Gastrointestinal dysmotility in Rett syndrome. *Journal of Pediatric Gastroenterology and Nutrition*, **2014**, 58, 237-44	2.8	36
33	Experience of gastrostomy using a quality care framework: the example of rett syndrome. *Medicine (United States)*, **2014**, 93, e328	1.8	19
32	Community participation for girls and women living with Rett syndrome. *Disability and Rehabilitation*, **2014**, 36, 894-9	2.4	12
31	Perspectives on hand function in girls and women with Rett syndrome. *Developmental Neurorehabilitation*, **2014**, 17, 210-7	1.8	14
30	Prevalence, clinical investigation, and management of gallbladder disease in Rett syndrome. *Developmental Medicine and Child Neurology*, **2014**, 56, 756-62	3.3	12
29	Twenty years of surveillance in Rett syndrome: what does this tell us?. *Orphanet Journal of Rare Diseases*, **2014**, 9, 87	4.2	72
28	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
27	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
26	Early development and regression in Rett syndrome. *Clinical Genetics*, **2013**, 84, 572-6	4	29
25	Caring for a child with severe intellectual disability in China: the example of Rett syndrome. *Disability and Rehabilitation*, **2013**, 35, 343-51	2.4	10
24	Assessment and management of nutrition and growth in Rett syndrome. *Journal of Pediatric Gastroenterology and Nutrition*, **2013**, 57, 451-60	2.8	37
23	Resourceful and creative methods are necessary to research rare disorders. *Developmental Medicine and Child Neurology*, **2013**, 55, 870-1	3.3	3
22	Using a large international sample to investigate epilepsy in Rett syndrome. *Developmental Medicine and Child Neurology*, **2013**, 55, 553-8	3.3	47
21	Barriers to diagnosis of a rare neurological disorder in China--lived experiences of Rett syndrome families. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1-9	2.5	13

20	Genetic drift. The diagnostic odyssey to Rett syndrome: the experience of an Australian family. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 10-2	2.5	12
19	Initial assessment of the StepWatch Activity Monitor to measure walking activity in Rett syndrome. *Disability and Rehabilitation*, **2012**, 34, 1010-5	2.4	14
18	The phenotype associated with a large deletion on MECP2. *European Journal of Human Genetics*, **2012**, 20, 921-7	5.3	25
17	The conductive environment enhances gross motor function of girls with Rett syndrome. A pilot study. *Developmental Neurorehabilitation*, **2012**, 15, 19-25	1.8	25
16	Use of equipment and respite services and caregiver health among Australian families living with Rett syndrome. *Research in Autism Spectrum Disorders*, **2011**, 5, 722-732	3	14
15	Trends in the diagnosis of Rett syndrome in Australia. *Pediatric Research*, **2011**, 70, 313-9	3.2	90
14	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
13	Altered attainment of developmental milestones influences the age of diagnosis of rett syndrome. *Journal of Child Neurology*, **2011**, 26, 980-7	2.5	33
12	Valproate and risk of fracture in Rett syndrome. *Archives of Disease in Childhood*, **2010**, 95, 444-8	2.2	29
11	Linking MECP2 and pain sensitivity: the example of Rett syndrome. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1197-205	2.5	62
10	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
9	Stereotypical hand movements in 144 subjects with Rett syndrome from the population-based Australian database. *Movement Disorders*, **2010**, 25, 282-8	7	42
8	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
7	Parental experiences of scoliosis management in Rett syndrome. *Disability and Rehabilitation*, **2009**, 31, 1917-24	2.4	10
6	Impact of scoliosis surgery on activities of daily living in females with Rett syndrome. *Journal of Pediatric Orthopaedics*, **2009**, 29, 369-74	2.4	14
5	Guidelines for management of scoliosis in Rett syndrome patients based on expert consensus and clinical evidence. *Spine*, **2009**, 34, E607-17	3.3	50
4	A guide for the assessment and management of vitamin D status in people with intellectual disability (developed as an AADDM Working Party initiative). *Journal of Intellectual and Developmental Disability*, **2008**, 33, 184-8	1.9	2
3	Early determinants of fractures in Rett syndrome. *Pediatrics*, **2008**, 121, 540-6	7.4	60

2	Gross motor profile in rett syndrome as determined by video analysis. *Neuropediatrics*, **2008**, 39, 205-10	1.6	47
1	Development of a video-based evaluation tool in Rett syndrome. *Journal of Autism and Developmental Disorders*, **2007**, 37, 1636-46	4.6	31