

# Jennepher Anne Downs

## List of Publications by Citations

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163  
papers

2,737  
citations

30  
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173  
ext. papers

3,512  
ext. citations

3.3  
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5.31  
L-index

#	Paper	IF	Citations
163	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
162	Clinical and biological progress over 50 years in Rett syndrome. <i>Nature Reviews Neurology</i> , <b>2017</b> , 13, 37-51	5.5	120
161	Trends in the diagnosis of Rett syndrome in Australia. <i>Pediatric Research</i> , <b>2011</b> , 70, 313-9	3.2	90
160	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
159	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
158	Early determinants of fractures in Rett syndrome. <i>Pediatrics</i> , <b>2008</b> , 121, 540-6	7.4	60
157	The trajectories of sleep disturbances in Rett syndrome. <i>Journal of Sleep Research</i> , <b>2015</b> , 24, 223-33	5.8	59
156	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
155	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
154	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
153	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
152	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
151	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
150	Gross motor profile in rett syndrome as determined by video analysis. <i>Neuropediatrics</i> , <b>2008</b> , 39, 205-10	1.6	47
149	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
148	Validating the Rett Syndrome Gross Motor Scale. <i>PLoS ONE</i> , <b>2016</b> , 11, e0147555	3.7	42
147	The Natural History of Scoliosis in Females With Rett Syndrome. <i>Spine</i> , <b>2016</b> , 41, 856-63	3.3	40

146	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
145	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
144	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
143	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
142	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
141	Gastrointestinal dysmotility in Rett syndrome. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2014</b> , 58, 237-44	2.8	36
140	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
139	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
138	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
137	Expanding the clinical picture of the MECP2 Duplication syndrome. <i>Clinical Genetics</i> , <b>2017</b> , 91, 557-563	4	32
136	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
135	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
134	Change in gross motor abilities of girls and women with rett syndrome over a 3- to 4-year period. <i>Journal of Child Neurology</i> , <b>2011</b> , 26, 1237-45	2.5	30
133	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
132	Early development and regression in Rett syndrome. <i>Clinical Genetics</i> , <b>2013</b> , 84, 572-6	4	29
131	Valproate and risk of fracture in Rett syndrome. <i>Archives of Disease in Childhood</i> , <b>2010</b> , 95, 444-8	2.2	29
130	variants: Improving our understanding of a rare neurologic disorder. <i>Neurology: Genetics</i> , <b>2017</b> , 3, e200	3.8	27
129	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

128	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
127	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
126	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
125	The conductive environment enhances gross motor function of girls with Rett syndrome. A pilot study. <i>Developmental Neurorehabilitation</i> , <b>2012</b> , 15, 19-25	1.8	25
124	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
123	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
122	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
121	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
120	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
119	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
118	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
117	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
116	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
115	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
114	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
113	Experience of gastrostomy using a quality care framework: the example of rett syndrome. <i>Medicine (United States)</i> , <b>2014</b> , 93, e328	1.8	19
112	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
111	Severity Assessment in CDKL5 Deficiency Disorder. <i>Pediatric Neurology</i> , <b>2019</b> , 97, 38-42	2.9	18

110	Very Early Identification and Intervention for Infants at Risk of Neurodevelopmental Disorders: A Transdiagnostic Approach. <i>Child Development Perspectives</i> , <b>2019</b> , 13, 97-103	5.5	17
109	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 Psychiatry</i> , <b>2020</b> , 59, 1036-1048	7.2	17
108	Quality of Life and Psychosocial Well-Being in Youth With Neuromuscular Disorders Who Are Wheelchair Users: A Systematic Review. <i>Archives of Physical Medicine and Rehabilitation</i> , <b>2017</b> , 98, 1004-1017.e17	2.8	17
107	"The problem with running"--comparing the propulsion strategy of children with developmental coordination disorder and typically developing children. <i>Gait and Posture</i> , <b>2014</b> , 39, 547-52	2.6	16
106	Impact of biobanks on research outcomes in rare diseases: a systematic review. <i>Orphanet Journal of Rare Diseases</i> , <b>2018</b> , 13, 202	4.2	16
105	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
104	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
103	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
102	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
101	Building the repertoire of measures of walking in Rett syndrome. <i>Disability and Rehabilitation</i> , <b>2017</b> , 39, 1926-1931	2.4	14
100	Perspectives on hand function in girls and women with Rett syndrome. <i>Developmental Neurorehabilitation</i> , <b>2014</b> , 17, 210-7	1.8	14
99	Initial assessment of the StepWatch Activity Monitor to measure walking activity in Rett syndrome. <i>Disability and Rehabilitation</i> , <b>2012</b> , 34, 1010-5	2.4	14
98	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
97	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
96	Addressing challenges in gaining informed consent for a research study investigating falls in people with intellectual disability. <i>British Journal of Learning Disabilities</i> , <b>2018</b> , 46, 92-100	1	13
95	Respiratory morbidity in Rett syndrome: an observational study. <i>Developmental Medicine and Child Neurology</i> , <b>2018</b> , 60, 951-957	3.3	13
94	Parent-reported health-related quality of life of children with Down syndrome: a descriptive study. <i>Developmental Medicine and Child Neurology</i> , <b>2018</b> , 60, 402-408	3.3	13
93	Barriers to diagnosis of a rare neurological disorder in China--lived experiences of Rett syndrome families. <i>American Journal of Medical Genetics, Part A</i> , <b>2012</b> , 158A, 1-9	2.5	13

92	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
91	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
90	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
89	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
88	Community participation for girls and women living with Rett syndrome. <i>Disability and Rehabilitation</i> , <b>2014</b> , 36, 894-9	2.4	12
87	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
86	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
85	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
84	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
83	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
82	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
81	Powered standing wheelchairs promote independence, health and community involvement in adolescents with Duchenne muscular dystrophy. <i>Neuromuscular Disorders</i> , <b>2019</b> , 29, 221-230	2.9	10
80	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
79	Parental experiences of scoliosis management in Rett syndrome. <i>Disability and Rehabilitation</i> , <b>2009</b> , 31, 1917-24	2.4	10
78	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
77	Incidence and prevalence of falls in adults with intellectual disability living in the community: a systematic review. <i>JBI Database of Systematic Reviews and Implementation Reports</i> , <b>2019</b> , 17, 390-413	1.6	10
76	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
75	A validation study of a modified Bouchard activity record that extends the concept of RiptimeRto Rett syndrome. <i>Developmental Medicine and Child Neurology</i> , <b>2015</b> , 57, 1137-42	3.3	9

74	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
73	Epidemiology of gastrostomy insertion for children and adolescents with intellectual disability. <i>European Journal of Pediatrics</i> , <b>2019</b> , 178, 351-361	4.1	8
72	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
71	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
70	Reliability of the Quality of Life Inventory-Disability Measure in Children with Intellectual Disability. <i>Journal of Developmental and Behavioral Pediatrics</i> , <b>2020</b> , 41, 534-539	2.4	7
69	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
68	Benefits of powered standing wheelchair devices for adolescents with Duchenne muscular dystrophy in the first year of use. <i>Journal of Paediatrics and Child Health</i> , <b>2020</b> , 56, 1419-1425	1.3	6
67	Family satisfaction following spinal fusion in Rett syndrome. <i>Developmental Neurorehabilitation</i> , <b>2016</b> , 19, 31-7	1.8	6
66	Measurement of Sedentary Behaviors or "Downtime" in Rett Syndrome. <i>Journal of Child Neurology</i> , <b>2017</b> , 32, 1009-1013	2.5	6
65	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
64	Propulsion strategy in the gait of primary school children; the effect of age and speed. <i>Human Movement Science</i> , <b>2016</b> , 50, 54-61	2.4	5
63	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
62	Mental wellbeing in non-ambulant youth with neuromuscular disorders: What makes the difference?. <i>Neuromuscular Disorders</i> , <b>2019</b> , 29, 48-58	2.9	5
61	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
60	Oral health experiences of individuals with Rett syndrome: a retrospective study. <i>BMC Oral Health</i> , <b>2018</b> , 18, 195	3.7	5
59	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
58	Feasibility and Effectiveness of an Individualized 12-Week "Uptime" Participation (U-PART) Intervention in Girls and Women With Rett Syndrome. <i>Physical Therapy</i> , <b>2020</b> , 100, 168-179	3.3	4
57	The Brain Basis of Comorbidity in Neurodevelopmental Disorders. <i>Current Developmental Disorders Reports</i> , <b>2019</b> , 6, 9-18	1.9	4

56	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
55	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
54	The development of a consensus statement for the prescription of powered wheelchair standing devices in Duchenne muscular dystrophy. <i>Disability and Rehabilitation</i> , <b>2020</b> , 1-9	2.4	4
53	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
52	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
51	Facilitators and Barriers of Participation in "Uptime" Activities in Girls and Women With Rett Syndrome: Perspectives From Parents and Professionals. <i>Qualitative Health Research</i> , <b>2019</b> , 29, 609-619	3.9	4
50	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
49	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
48	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
47	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. <i>BMC Geriatrics</i> , <b>2018</b> , 18, 171	4.1	3
46	Incidence and associated risk factors for falls in adults with intellectual disability. <i>Journal of Intellectual Disability Research</i> , <b>2019</b> , 63, 1441-1452	3.2	3
45	Incidence and prevalence of falls in adults with intellectual disability living in the community: a systematic review protocol. <i>JBIM Database of Systematic Reviews and Implementation Reports</i> , <b>2017</b> , 15, 1819-1823	1.6	3
44	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
43	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
42	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
41	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
40	A Pilot Study Delivering Physiotherapy Support for Rett Syndrome Using a Telehealth Framework Suitable for COVID-19 Lockdown. <i>Developmental Neurorehabilitation</i> , <b>2021</b> , 24, 429-434	1.8	3
39	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



38	Women Diagnosed with Ovarian Cancer: Patient and Carer Experiences and Perspectives. <i>Patient Related Outcome Measures</i> , <b>2021</b> , 12, 33-43	2.9	3
37	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
36	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
35	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
34	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. <i>JMIR Research Protocols</i> , <b>2020</b> , 9, e19803	2	2
33	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
32	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
31	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
30	Determinants of sleep problems in children with intellectual disability. <i>Journal of Sleep Research</i> , <b>2021</b> , 30, e13361	5.8	2
29	Caregiver-mediated interventions to support self-regulation among infants and young children (0-5 years): a protocol for a realist review. <i>BMJ Open</i> , <b>2021</b> , 11, e046078	3	2
28	A fine balance and a shared learning journey: Exploring healthcare engagement through the experiences of youth with Neuromuscular Disorders. <i>NeuroRehabilitation</i> , <b>2016</b> , 39, 519-534	2	2
27	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
26	Early Motor Function of Children With Autism Spectrum Disorder: A Systematic Review. <i>Pediatrics</i> , <b>2021</b> , 147,	7.4	2
25	Exploring enablers and barriers to accessing health services after a fall among people with intellectual disability. <i>Journal of Applied Research in Intellectual Disabilities</i> , <b>2020</b> , 33, 604-617	2.2	1
24	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
23	Using a trauma informed practice framework to enhance understanding of and identify support strategies for behavioural difficulties in young people with Prader-Willi syndrome. <i>Research in Developmental Disabilities</i> , <b>2021</b> , 110, 103839	2.7	1
22	Parent Carer Quality of Life and Night-Time Attendance in Non-Ambulant Youth with Neuromuscular Disorders. <i>Developmental Neurorehabilitation</i> , <b>2021</b> , 24, 456-465	1.8	1
21	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

20	How can clinical ethics guide the management of comorbidities in the child with Rett syndrome? <i>Journal of Paediatrics and Child Health</i> , <b>2016</b> , 52, 809-13	1.3	1
19	Sleep-disordered breathing in Australian children with Prader-Willi syndrome following initiation of growth hormone therapy. <i>Journal of Paediatrics and Child Health</i> , <b>2021</b> ,	1.3	1
18	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
17	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
16	Medical Comorbidities in MECP2 Duplication Syndrome: Results from the International MECP2 Duplication Database. <i>Children</i> , <b>2022</b> , 9, 633	2.8	1
15	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 <sup>3.7</sup>	3.7	0
14	Survival of children and adolescents with intellectual disability following gastrostomy insertion. <i>Journal of Intellectual Disability Research</i> , <b>2020</b> , 64, 497-511	3.2	0
13	Decline in gross motor skills in adult Rett syndrome; results from a Danish longitudinal study. <i>American Journal of Medical Genetics, Part A</i> , <b>2021</b> , 185, 3683-3693	2.5	0
12	The Rett Syndrome Gross Motor Scale - Dutch Version (RSGMS-NL) Can Reliably Assess Gross Motor Skills in Dutch Individuals with Rett Syndrome. <i>Developmental Neurorehabilitation</i> , <b>2021</b> , 1-7	1.8	0
11	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
10	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	0
9	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
8	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
7	Genetic and epigenetic influences on the phenotype of Rett syndrome <b>2019</b> , 183-217		
6	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	
5	Parent and therapist perspectives on "uptime" activities and participation in Rett syndrome. <i>Disability and Rehabilitation</i> , <b>2021</b> , 1-8	2.4	
4	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	
3	Family-Centered Telehealth Supporting Motor Skills and Activity in Individuals With Rett Syndrome. <i>Advances in Medical Technologies and Clinical Practice Book Series</i> , <b>2022</b> , 147-171	0.3	

- 2 The Lived Experience of Parents Receiving the Diagnosis of CDKL5 Deficiency Disorder for Their Child.. *Journal of Child Neurology*, **2022**, 8830738221076285 2.5
- 1 Daytime sleepiness and emotional and behavioral disturbances in Prader-Willi syndrome.. *European Journal of Pediatrics*, **2022**, 1 4.1