Jennepher Anne Downs

List of Publications by Year
in descending order

170 papers	4,280 citations	109137 35 h-index	174990 52 g-index
173 all docs	173 docs citations	173 times ranked	3008 citing authors

#	Article	IF	Citations
1	The CDKL5 disorder is an independent clinical entity associated with early-onset encephalopathy. European Journal of Human Genetics, 2013, 21, 266-273.	1.4	220
2	Clinical and biological progress over 50 years in Rett syndrome. Nature Reviews Neurology, 2017, 13, 37-51.	4.9	155
3	Trends in the Diagnosis of Rett Syndrome in Australia. Pediatric Research, 2011, 70, 313-319.	1.1	119
4	Twenty years of surveillance in Rett syndrome: what does this tell us?. Orphanet Journal of Rare Diseases, 2014, 9, 87.	1.2	98
5	The prevalence of mental health disorders and symptoms in children and adolescents with cerebral palsy: a systematic review and metaâ€analysis. Developmental Medicine and Child Neurology, 2018, 60, 30-38.	1.1	84
6	The trajectories of sleep disturbances in Rett syndrome. Journal of Sleep Research, 2015, 24, 223-233.	1.7	83
7	Prevalence and onset of comorbidities in the CDKL5 disorder differ from Rett syndrome. Orphanet Journal of Rare Diseases, 2016, 11, 39.	1.2	81
8	Linking MECP2 and pain sensitivity: The example of Rett syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 1197-1205.	0.7	80
9	There is variability in the attainment of developmental milestones in the CDKL5 disorder. Journal of Neurodevelopmental Disorders, 2015, 7, 2.	1.5	74
10	Seizure variables and their relationship to genotype and functional abilities in the CDKL5 disorder. Neurology, 2016, 87, 2206-2213.	1.5	74
11	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.	1.3	71
12	Early Determinants of Fractures in Rett Syndrome. Pediatrics, 2008, 121, 540-546.	1.0	67
13	Guidelines for Management of Scoliosis in Rett Syndrome Patients Based on Expert Consensus and Clinical Evidence. Spine, 2009, 34, E607-E617.	1.0	65
14	Functional abilities in children and adults with the CDKL5 disorder. American Journal of Medical Genetics, Part A, 2016, 170, 2860-2869.	0.7	65
15	Transition to adulthood for young people with intellectual disability: the experiences of their families. European Child and Adolescent Psychiatry, 2016, 25, 1369-1381.	2.8	59
16	Stereotypical hand movements in 144 subjects with Rett syndrome from the populationâ€based Australian database. Movement Disorders, 2010, 25, 282-288.	2.2	52
17	Using a large international sample to investigate epilepsy in <scp>R</scp>ett syndrome. Developmental Medicine and Child Neurology, 2013, 55, 553-558.	1.1	52
18	Conceptualizing a quality of life framework for girls with Rett syndrome using qualitative methods. American Journal of Medical Genetics, Part A, 2016, 170, 645-653.	0.7	52

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19	CDKL5 variants. Neurology: Genetics, 2017, 3, e200.	0.9	52
20	Gross Motor Profile in Rett Syndrome as Determined by Video Analysis. Neuropediatrics, 2008, 39, 205-210.	0.3	51
21	Validating the Rett Syndrome Gross Motor Scale. PLoS ONE, 2016, 11, e0147555.	1.1	51
22	Use of the ketogenic diet to manage refractory epilepsy in <scp>CDKL</scp>5 disorder: Experience of >100 patients. Epilepsia, 2017, 58, 1415-1422.	2.6	51
23	Environmental enrichment intervention for Rett syndrome: an individually randomised stepped wedge trial. Orphanet Journal of Rare Diseases, 2018, 13, 3.	1.2	51
24	The Natural History of Scoliosis in Females With Rett Syndrome. Spine, 2016, 41, 856-863.	1.0	50
25	Assessment and Management of Nutrition and Growth in Rett Syndrome. Journal of Pediatric Gastroenterology and Nutrition, 2013, 57, 451-460.	0.9	48
26	Gastrointestinal Dysmotility in Rett Syndrome. Journal of Pediatric Gastroenterology and Nutrition, 2014, 58, 237-244.	0.9	48
27	Psychometric properties of the Quality of Life Inventory-Disability (QI-Disability) measure. Quality of Life Research, 2019, 28, 783-794.	1.5	48
28	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.	1.2	46
29	Level of purposeful hand function as a marker of clinical severity in Rett syndrome. Developmental Medicine and Child Neurology, 2010, 52, 817-823.	1.1	45
30	Expanding the clinical picture of the MECP2 Duplication syndrome. Clinical Genetics, 2017, 91, 557-563.	1.0	45
31	Clinical Guidelines for Management of Bone Health in Rett Syndrome Based on Expert Consensus and Available Evidence. PLoS ONE, 2016, 11, e0146824.	1.1	45
32	CDKL5 deficiency disorder: clinical features, diagnosis, and management. Lancet Neurology, The, 2022, 21, 563-576.	4.9	44
33	Severity Assessment in CDKL5 Deficiency Disorder. Pediatric Neurology, 2019, 97, 38-42.	1.0	43
34	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.	0.8	42
35	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.	0.3	40
36	Functioning, participation, and quality of life in children with intellectual disability: an observational study. Developmental Medicine and Child Neurology, 2021, 63, 89-96.	1.1	40

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37	Determinants of sleep disturbances in Rett syndrome: Novel findings in relation to genotype. American Journal of Medical Genetics, Part A, 2016, 170, 2292-2300.	0.7	38
38	Altered Attainment of Developmental Milestones Influences the Age of Diagnosis of Rett Syndrome. Journal of Child Neurology, 2011, 26, 980-987.	0.7	37
39	Early development and regression in Rett syndrome. Clinical Genetics, 2013, 84, 572-576.	1.0	37
40	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.	0.6	36
41	Development of a Video-based Evaluation Tool in Rett Syndrome. Journal of Autism and Developmental Disorders, 2007, 37, 1636-1646.	1.7	35
42	Valproate and risk of fracture in Rett syndrome. Archives of Disease in Childhood, 2010, 95, 444-448.	1.0	35
43	Very Early Identification and Intervention for Infants at Risk of Neurodevelopmental Disorders: AÂTransdiagnostic Approach. Child Development Perspectives, 2019, 13, 97-103.	2.1	34
44	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, 1237-1245.	0.7	33
45	Parental perspectives on the communication abilities of their daughters with Rett syndrome. Developmental Neurorehabilitation, 2016, 19, 17-25.	0.5	33
46	Vagus nerve stimulation for the treatment of refractory epilepsy in the CDKL5 Deficiency Disorder. Epilepsy Research, 2018, 146, 36-40.	0.8	33
47	Autonomic breathing abnormalities in Rett syndrome: caregiver perspectives in an international database study. Journal of Neurodevelopmental Disorders, 2017, 9, 15.	1.5	32
48	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, 2535-2542.	0.7	31
49	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.	0.7	31
50	The phenotype associated with a large deletion on MECP2. European Journal of Human Genetics, 2012, 20, 921-927.	1.4	30
51	Aspects of speechâ€language abilities are influenced by MECP2 mutation type in girls with Rett syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 354-362.	0.7	30
52	Impact of biobanks on research outcomes in rare diseases: a systematic review. Orphanet Journal of Rare Diseases, 2018, 13, 202.	1.2	30
53	The conductive environment enhances gross motor function of girls with Rett syndrome. A pilot study. Developmental Neurorehabilitation, 2012, 15, 19-25.	0.5	28
54	Rett syndrome: establishing a novel outcome measure for walking activity in an era of clinical trials for rare disorders. Disability and Rehabilitation, 2015, 37, 1992-1996.	0.9	28

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55	Evolving Trends of Gastrostomy Insertion Within a Pediatric Population. Journal of Pediatric Gastroenterology and Nutrition, 2018, 67, e89-e94.	0.9	28
56	The incidence, prevalence and clinical features of MECP2 duplication syndrome in Australian children. Journal of Paediatrics and Child Health, 2019, 55, 1315-1322.	0.4	28
57	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.	2.8	28
58	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.	1.7	26
59	Relationship between family quality of life and day occupations of young people with Down syndrome. Social Psychiatry and Psychiatric Epidemiology, 2014, 49, 1455-1465.	1.6	25
60	Quantification of walkingâ€based physical activity and sedentary time in individuals with Rett syndrome. Developmental Medicine and Child Neurology, 2017, 59, 605-611.	1.1	25
61	Surgical fusion of early onset severe scoliosis increases survival in Rett syndrome: a cohort study. Developmental Medicine and Child Neurology, 2016, 58, 632-638.	1.1	24
62	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.	0.5	24
63	Data Linkage: Canadian and Australian Perspectives on a Valuable Methodology for Intellectual and Developmental Disability Research. Intellectual and Developmental Disabilities, 2019, 57, 439-462.	0.6	23
64	Exploring quality of life in individuals with a severe developmental and epileptic encephalopathy, CDKL5 Deficiency Disorder. Epilepsy Research, 2021, 169, 106521.	0.8	23
65	Experience of Gastrostomy Using a Quality Care Framework. Medicine (United States), 2014, 93, e328.	0.4	22
66	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.	1.7	22
67	Early Motor Function of Children With Autism Spectrum Disorder: A Systematic Review. Pediatrics, 2021, 147, .	1.0	22
68	Perspectives on hand function in girls and women with Rett syndrome. Developmental Neurorehabilitation, 2014, 17, 210-217.	0.5	21
69	Parentâ€reported healthâ€related quality of life of children with Down syndrome: a descriptive study. Developmental Medicine and Child Neurology, 2018, 60, 402-408.	1.1	21
70	Content validation of the Quality of Life Inventoryâ€”Disability. Child: Care, Health and Development, 2019, 45, 654-659.	0.8	21
71	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.	0.7	20
72	Building the repertoire of measures of walking in Rett syndrome. Disability and Rehabilitation, 2017, 39, 1926-1931.	0.9	20

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73	Respiratory morbidity in Rett syndrome: an observational study. Developmental Medicine and Child Neurology, 2018, 60, 951-957.	1.1	20
74	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.	2.2	20
75	Cannabis for refractory epilepsy in children: A review focusing on CDKL5 Deficiency Disorder. Epilepsy Research, 2019, 151, 31-39.	0.8	20
76	Parent-observed thematic data on quality of life in children with autism spectrum disorder. Autism, 2019, 23, 71-80.	2.4	20
77	A brief history of MECP2 duplication syndrome: 20-years of clinical understanding. Orphanet Journal of Rare Diseases, 2022, 17, 131.	1.2	20
78	â€œThe problem with runningâ€â€”Comparing the propulsion strategy of children with Developmental Coordination Disorder and typically developing children. Gait and Posture, 2014, 39, 547-552.	0.6	18
79	Quantitative and qualitative insights into the experiences of children with Rett syndrome and their families. Wiener Medizinische Wochenschrift, 2016, 166, 338-345.	0.5	18
80	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.	0.8	18
81	Exploring genotypeâ€phenotype relationships in the <scp>CDKL5</scp> deficiency disorder using an international dataset. Clinical Genetics, 2021, 99, 157-165.	1.0	18
82	International Consensus Recommendations for the Assessment and Management of Individuals With CDKL5 Deficiency Disorder. Frontiers in Neurology, 0, 13, .	1.1	18
83	Impact of Scoliosis Surgery on Activities of Daily Living in Females With Rett Syndrome. Journal of Pediatric Orthopaedics, 2009, 29, 369-374.	0.6	16
84	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.	0.8	16
85	Initial assessment of the StepWatch Activity Monitorâ„¢ to measure walking activity in Rett syndrome. Disability and Rehabilitation, 2012, 34, 1010-1015.	0.9	16
86	Powered standing wheelchairs promote independence, health and community involvement in adolescents with Duchenne muscular dystrophy. Neuromuscular Disorders, 2019, 29, 221-230.	0.3	16
87	Epidemiology of gastrostomy insertion for children and adolescents with intellectual disability. European Journal of Pediatrics, 2019, 178, 351-361.	1.3	16
88	Reliability of the Quality of Life Inventory-Disability Measure in Children with Intellectual Disability. Journal of Developmental and Behavioral Pediatrics, 2020, 41, 534-539.	0.6	16
89	Barriers to diagnosis of a rare neurological disorder in Chinaâ€”Lived experiences of Rett syndrome families. American Journal of Medical Genetics, Part A, 2012, 158A, 1-9.	0.7	15
90	Comorbidities and quality of life in children with intellectual disability. Child: Care, Health and Development, 2021, 47, 654-666.	0.8	15

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91	A validation study of a modified Bouchard activity record that extends the concept of â€˜uptimeâ€™ to Rett syndrome. Developmental Medicine and Child Neurology, 2015, 57, 1137-1142.	1.1	14
92	The diagnostic odyssey to Rett syndrome: The experience of an Australian family. American Journal of Medical Genetics, Part A, 2012, 158A, 10-12.	0.7	13
93	Caring for a child with severe intellectual disability in China: The example of Rett syndrome. Disability and Rehabilitation, 2013, 35, 343-351.	0.9	13
94	Community participation for girls and women living with Rett syndrome. Disability and Rehabilitation, 2014, 36, 894-899.	0.9	13
95	Prevalence, clinical investigation, and management of gallbladder disease in Rett syndrome. Developmental Medicine and Child Neurology, 2014, 56, 756-762.	1.1	13
96	Spinal fusion in girls with <scp>R</scp>ett syndrome: postâ€operative recovery and family experiences. Child: Care, Health and Development, 2015, 41, 1000-1009.	0.8	13
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.	0.7	13
98	Risk of Developmental Disorders in Children of Immigrant Mothers: A Population-Based Data Linkage Evaluation. Journal of Pediatrics, 2019, 204, 275-284.e3.	0.9	13
99	Cyclin-dependentâ€“like kinase 5 is required for pain signaling in human sensory neurons and mouse models. Science Translational Medicine, 2020, 12, .	5.8	13
100	Women Diagnosed with Ovarian Cancer: Patient and Carer Experiences and Perspectives. Patient Related Outcome Measures, 2021, Volume 12, 33-43.	0.7	13
101	A Pilot Study Delivering Physiotherapy Support for Rett Syndrome Using a Telehealth Framework Suitable for COVID-19 Lockdown. Developmental Neurorehabilitation, 2021, 24, 1-6.	0.5	13
102	Longitudinal bone mineral content and density in Rett syndrome and their contributing factors. Bone, 2015, 74, 191-198.	1.4	12
103	Determinants of sleep problems in children with intellectual disability. Journal of Sleep Research, 2021, 30, e13361.	1.7	12
104	Parental experiences of scoliosis management in Rett syndrome. Disability and Rehabilitation, 2009, 31, 1917-1924.	0.9	11
105	Measurement of Sedentary Behaviors or â€œDowntimeâ€•in Rett Syndrome. Journal of Child Neurology, 2017, 32, 1009-1013.	0.7	11
106	Choice making in Rett syndrome: a descriptive study using video data. Disability and Rehabilitation, 2018, 40, 813-819.	0.9	11
107	Requirements for improving health and wellâ€being of children with Praderâ€Willi syndrome and their families. Journal of Paediatrics and Child Health, 2019, 55, 1029-1037.	0.4	11
108	Feasibility and Effectiveness of an Individualized 12-Week â€œUptimeâ€•Participation (U-PART) Intervention in Girls and Women With Rett Syndrome. Physical Therapy, 2020, 100, 168-179.	1.1	11

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109	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.	0.9	11
110	Expanding the phenotype of the CDKL5 deficiency disorder: Are seizures mandatory?. American Journal of Medical Genetics, Part A, 2020, 182, 1217-1222.	0.7	11
111	Content Validation of Clinician-Reported Items for a Severity Measure for CDKL5 Deficiency Disorder. Journal of Child Neurology, 2021, 36, 998-1006.	0.7	11
112	Oral health experiences of individuals with Rett syndrome: a retrospective study. BMC Oral Health, 2018, 18, 195.	0.8	10
113	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.	0.9	10
114	Modifiable child and caregiver factors that influence community participation among children with Down syndrome. Disability and Rehabilitation, 2022, 44, 600-607.	0.9	10
115	Assessment of a Clinical Trial Metric for Rett Syndrome: Critical Analysis of the Rett Syndrome Behaviour Questionnaire. Pediatric Neurology, 2020, 111, 4.	1.0	10
116	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.	0.4	10
117	Sleepâ€disordered breathing in Australian children with Praderâ€Willi syndrome following initiation of growth hormone therapy. Journal of Paediatrics and Child Health, 2022, 58, 248-255.	0.4	10
118	Influences on the trajectory and subsequent outcomes in CDKL5 deficiency disorder. Epilepsia, 2022, 63, 352-363.	2.6	10
119	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.	0.6	9
120	Improving clinical trial readiness to accelerate development of new therapeutics for Rett syndrome. Orphanet Journal of Rare Diseases, 2022, 17, 108.	1.2	9
121	Incidence and associated risk factors for falls in adults with intellectual disability. Journal of Intellectual Disability Research, 2019, 63, 1441-1452.	1.2	8
122	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.	0.9	8
123	Mental wellbeing in non-ambulant youth with neuromuscular disorders: What makes the difference?. Neuromuscular Disorders, 2019, 29, 48-58.	0.3	8
124	Risk of Hospitalizations Following Gastrostomy in Children with Intellectual Disability. Journal of Pediatrics, 2020, 217, 131-138.e10.	0.9	8
125	Propulsion strategy in the gait of primary school children; the effect of age and speed. Human Movement Science, 2016, 50, 54-61.	0.6	7
126	Family satisfaction following spinal fusion in Rett syndrome. Developmental Neurorehabilitation, 2016, 19, 31-37.	0.5	7

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127	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.	1.7	7
128	Hospital admissions in children with developmental disabilities from ethnic minority backgrounds. Developmental Medicine and Child Neurology, 2020, 62, 470-476.	1.1	7
129	The development of a consensus statement for the prescription of powered wheelchair standing devices in Duchenne muscular dystrophy. Disability and Rehabilitation, 2022, 44, 1889-1897.	0.9	7
130	Determinants of quality of life in Rett syndrome: new findings on associations with genotype. Journal of Medical Genetics, 2021, 58, 637-644.	1.5	7
131	Gastrostomy and quality of life in children with intellectual disability: a qualitative study. Archives of Disease in Childhood, 2020, 105, 969-974.	1.0	7
132	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.	0.7	7
133	The perceived effects of cannabis products in the management of seizures in CDKL5 Deficiency Disorder. Epilepsy and Behavior, 2021, 122, 108152.	0.9	7
134	A fine balance and a shared learning journey: Exploring healthcare engagement through the experiences of youth with Neuromuscular Disorders. NeuroRehabilitation, 2016, 39, 519-534.	0.5	6
135	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.	0.9	6
136	The Brain Basis of Comorbidity in Neurodevelopmental Disorders. Current Developmental Disorders Reports, 2019, 6, 9-18.	0.9	6
137	Facilitators and Barriers of Participation in â€œUptimeâ€•Activities in Girls and Women With Rett Syndrome: Perspectives From Parents and Professionals. Qualitative Health Research, 2019, 29, 609-619.	1.0	6
138	A preliminary investigation of the effects of prenatal alcohol exposure on facial morphology in children with Autism Spectrum Disorder. Alcohol, 2020, 86, 75-80.	0.8	6
139	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.	0.5	6
140	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, 184-188.	1.1	5
141	A qualitative investigation of recovery after femoral fracture in Rett syndrome. Child: Care, Health and Development, 2017, 43, 232-239.	0.8	5
142	Oral health care and service utilisation in individuals with Rett syndrome: an international crossâ€sectional study. Journal of Intellectual Disability Research, 2021, 65, 561-576.	1.2	5
143	Modelling quality of life in children with intellectual disability using regression trees. Developmental Medicine and Child Neurology, 2022, 64, 1145-1155.	1.1	5
144	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.	0.8	4

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145	Survival of children and adolescents with intellectual disability following gastrostomy insertion. Journal of Intellectual Disability Research, 2020, 64, 497-511.	1.2	4
146	Parent Carer Quality of Life and Night-Time Attendance in Non-Ambulant Youth with Neuromuscular Disorders. Developmental Neurorehabilitation, 2021, 24, 456-465.	0.5	4
147	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.	1.5	4
148	Implementing telehealth support to increase physical activity in girls and women with Rett syndromeâ€"ActivRett: protocol for a waitlist randomised controlled trial. BMJ Open, 2020, 10, e042446.	0.8	4
149	The Lived Experience of Parentsâ€™ Receiving the Diagnosis of CDKL5 Deficiency Disorder for Their Child. Journal of Child Neurology, 2022, 37, 451-460.	0.7	4
150	Initial Validation and Reliability of the CDKL5 Deficiency Disorder Hand Function Scale (CDD-Hand). Journal of Child Neurology, 2022, 37, 541-547.	0.7	4
151	Resourceful and creative methods are necessary to research rare disorders. Developmental Medicine and Child Neurology, 2013, 55, 870-871.	1.1	3
152	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.	1.7	3
153	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.	1.1	3
154	Oral health education and promotion in special needs children: Systematic review and metaâ€analysis. Oral Diseases, 2022, 28, 66-75.	1.5	3
155	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.	1.2	3
156	Longitudinal Evaluation of the Stability of Hand Function in Rett Syndrome. Journal of Pediatrics, 2021, 237, 244-249.e3.	0.9	3
157	Enablers and barriers in dental attendance in Rett syndrome: an international observational study. Special Care in Dentistry, 2022, , .	0.4	3
158	Negative impact of insomnia and daytime sleepiness on quality of life in individuals with the cyclinâ€dependent kinaseâ€like 5 deficiency disorder. Journal of Sleep Research, 2022, , e13600.	1.7	3
159	Medical Comorbidities in MECP2 Duplication Syndrome: Results from the International MECP2 Duplication Database. Children, 2022, 9, 633.	0.6	3
160	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.	1.3	2
161	Caregiver-mediated interventions to support self-regulation among infants and young children (0â€“5) Tj ETQq1 1 0.784314 rgBT /Ov	0.8	2
162	The Rett Syndrome Gross Motor Scale â€“ Dutch Version (RSGMS-NL) Can Reliably Assess Gross Motor Skills in Dutch Individuals with Rett Syndrome. Developmental Neurorehabilitation, 2022, 25, 133-139.	0.5	2

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163	How can clinical ethics guide the management of comorbidities in the child with Rett syndrome?. Journal of Paediatrics and Child Health, 2016, 52, 809-813.	0.4	1
164	Genetic and epigenetic influences on the phenotype of Rett syndrome. , 2019, , 183-217.		1
165	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.	1.7	1
166	Ketogenic diet to manage refractory epilepsy in the cdkl5 disorder. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, e1.27-e1.	0.9	0
167	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	0
168	Parent and therapist perspectives on "uptime" activities and participation in Rett syndrome. Disability and Rehabilitation, 2021, , 1-8.	0.9	0
169	Strengths and challenging behaviors in children and adolescents with<scp>Praderâ€Willi</scp>syndrome: Two sides to the coin. American Journal of Medical Genetics, Part A, 2022, 188, 1488-1496.	0.7	0
170	Daytime sleepiness and emotional and behavioral disturbances in Prader-Willi syndrome. European Journal of Pediatrics, 2022, , 1.	1.3	0