

Oliver W J Quarrell

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/4224337/publications.pdf>

Version: 2024-02-01

22
papers

567
citations

759233

12
h-index

713466

21
g-index

23
all docs

23
docs citations

23
times ranked

936
citing authors

#	ARTICLE	IF	CITATIONS
1	Pathogenetics of alveolar capillary dysplasia with misalignment of pulmonary veins. <i>Human Genetics</i> , 2016, 135, 569-586.	3.8	85
2	Managing juvenile Huntington's disease. <i>Neurodegenerative Disease Management</i> , 2013, 3, 267-276.	2.2	78
3	22 Years of predictive testing for Huntington's disease: the experience of the UK Huntington's Prediction Consortium. <i>European Journal of Human Genetics</i> , 2016, 24, 1396-1402.	2.8	73
4	Reduced penetrance alleles for Huntington's disease: a multi-centre direct observational study. <i>Journal of Medical Genetics</i> , 2006, 44, e68-e68.	3.2	67
5	Diagnostic genetic testing for Huntington's disease. <i>Practical Neurology</i> , 2015, 15, 80-84.	1.1	44
6	Task-Specific Training in Huntington Disease: A Randomized Controlled Feasibility Trial. <i>Physical Therapy</i> , 2014, 94, 1555-1568.	2.4	37
7	Defining the breakpoints of proximal chromosome 14q rearrangements in nine patients using flow-sorted chromosomes. <i>American Journal of Medical Genetics Part A</i> , 2001, 102, 173-182.	2.4	33
8	A survey-based study identifies common but unrecognized symptoms in a large series of juvenile Huntington's disease. <i>Neurodegenerative Disease Management</i> , 2017, 7, 307-315.	2.2	25
9	Discrepancies in reporting the CAG repeat lengths for Huntington's disease. <i>European Journal of Human Genetics</i> , 2012, 20, 20-26.	2.8	20
10	Homozygous <i>GDF2</i> nonsense mutations result in a loss of circulating BMP9 and BMP10 and are associated with either PAH or an "HHT-like" syndrome in children. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1685.	1.2	19
11	Large-scale open-source three-dimensional growth curves for clinical facial assessment and objective description of facial dysmorphism. <i>Scientific Reports</i> , 2021, 11, 12175.	3.3	17
12	Defining pediatric huntington disease: Time to abandon the term "Juvenile Huntington Disease"?. <i>Movement Disorders</i> , 2019, 34, 584-585.	3.9	16
13	Characterization of Gastric Mucosa Biopsies Reveals Alterations in Huntington's Disease. <i>PLOS Currents</i> , 2015, 7, .	1.4	10
14	Seizure frequency in adults with Wolf-Hirschhorn syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 2528-2531.	1.2	9
15	A hypomorphic allele of <i>SLC35D1</i> results in Schneckenbecken-like dysplasia. <i>Human Molecular Genetics</i> , 2019, 28, 3543-3551.	2.9	9
16	27 years of prenatal diagnosis for Huntington disease in the United Kingdom. <i>Genetics in Medicine</i> , 2019, 21, 1639-1643.	2.4	9
17	Assessment of the Performance of a Modified Motor Scale as Applied to Juvenile Onset Huntington's Disease. <i>Journal of Huntington's Disease</i> , 2019, 8, 181-193.	1.9	6
18	A case-note review of continued pregnancies found to be at a high risk of Huntington's disease: considerations for clinical practice. <i>European Journal of Human Genetics</i> , 2019, 27, 1215-1224.	2.8	4

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19	Letter in Response to Tibben et al., Risk Assessment for Huntington's Disease for (Future) Offspring Requires Offering Preconceptional CAG Analysis to Both Partners. <i>Journal of Huntington's Disease</i> , 2019, 8, 357-359.	1.9	2
20	Predictive genetic testing for Motor neuron disease: time for a guideline?. <i>European Journal of Human Genetics</i> , 2022, 30, 635-636.	2.8	2
21	Coinheritance of 2 New Potentially Damaging Heterozygous COL7A1 Variants in a Family With Autosomal Dominant Epidermolysis Bullosa Pruriginosa. <i>Pediatric and Developmental Pathology</i> , 2018, 21, 580-584.	1.0	1
22	Predictive testing of minors for Huntington's disease: The UK and Netherlands experiences. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2018, 177, 35-39.	1.7	1