VÃ©ronique David

List of Publications by Year
in descending order

29	1,663	304743 22	454955 30
papers	citations	h-index	g-index

31	31	31	1693
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Holoprosencephaly. Orphanet Journal of Rare Diseases, 2007, 2, 8.	2.7	299
2	Analysis of genotypeâ€“phenotype correlations in human holoprosencephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 133-141.	1.6	139
3	Molecular screening ofSHH,ZIC2,SIX3, andTGIF genes in patients with features of holoprosencephaly spectrum: Mutation review and genotype-phenotype correlations. Human Mutation, 2004, 24, 43-51.	2.5	128
4	New findings for phenotype-genotype correlations in a large European series of holoprosencephaly cases. Journal of Medical Genetics, 2011, 48, 752-760.	3.2	90
5	Twelve new patients with 13q deletion syndrome: Genotypeâ€“phenotype analyses in progress. European Journal of Medical Genetics, 2009, 52, 41-46.	1.3	80
6	The mutational spectrum of holoprosencephaly-associated changes within theSHHgene in humans predicts loss-of-function through either key structural alterations of the ligand or its altered synthesis. Human Mutation, 2009, 30, E921-E935.	2.5	77
7	Genotypic and phenotypic analysis of 396 individuals with mutations inSonic Hedgehog. Journal of Medical Genetics, 2012, 49, 473-479.	3.2	67
8	Current recommendations for the molecular evaluation of newly diagnosed holoprosencephaly patients. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 93-101.	1.6	62
9	Phenotypic and molecular variability of the holoprosencephalic spectrum. American Journal of Medical Genetics, Part A, 2004, 129A, 21-24.	1.2	61
10	The full spectrum of holoprosencephaly-associated mutations within theZIC2gene in humans predicts loss-of-function as the predominant disease mechanism. Human Mutation, 2009, 30, E541-E554.	2.5	56
11	Mutational Spectrum in Holoprosencephaly Shows That FGF is a New Major Signaling Pathway. Human Mutation, 2016, 37, 1329-1339.	2.5	56
12	Molecular evaluation of foetuses with holoprosencephaly shows high incidence of microdeletions in the HPE genes. Human Genetics, 2006, 119, 1-8.	3.8	52
13	NOTCH, a new signaling pathway implicated in holoprosencephaly. Human Molecular Genetics, 2011, 20, 1122-1131.	2.9	47
14	Array-CGH analysis indicates a high prevalence of genomic rearrangements in holoprosencephaly: An updated map of candidate loci. Human Mutation, 2009, 30, 1175-1182.	2.5	46
15	Holoprosencephaly: An update on cytogenetic abnormalities. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 86-92.	1.6	46
16	Integrated clinical and omics approach to rare diseases: novel genes and oligogenic inheritance in holoprosencephaly. Brain, 2019, 142, 35-49.	7.6	44
17	Molecular screening of the TGIF gene in holoprosencephaly: identification of two novel mutations. Human Genetics, 2003, 112, 131-134.	3.8	43
18	Recent advances in understanding inheritance of holoprosencephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 258-269.	1.6	42

#	ARTICLE	IF	CITATIONS
19	Homozygous STIL Mutation Causes Holoprosencephaly and Microcephaly in Two Siblings. PLoS ONE, 2015, 10, e0117418.	2.5	34
20	Identification of gene copy number variations in patients with mental retardation using array-CGH: Novel syndromes in a large French series. European Journal of Medical Genetics, 2010, 53, 66-75.	1.3	29
21	Novel genes upregulated when NOTCH signalling is disrupted during hypothalamic development. Neural Development, 2013, 8, 25.	2.4	26
22	MLPA screening reveals novel subtelomeric rearrangements in holoprosencephaly. Human Mutation, 2007, 28, 1189-1197.	2.5	25
23	NODAL and SHH dose-dependent double inhibition promotes an HPE-like phenotype in chick embryos. DMM Disease Models and Mechanisms, 2013, 6, 537-43.	2.4	20
24	A de novo variant in ADGRL2 suggests a novel mechanism underlying the previously undescribed association of extreme microcephaly with severely reduced sulcation and rhombencephalosynapsis. Acta Neuropathologica Communications, 2018, 6, 109.	5.2	20
25	Dynamic expression of Notch-dependent neurogenic markers in the chick embryonic nervous system. Frontiers in Neuroanatomy, 2014, 8, 158.	1.7	16
26	Clinical utility gene card for: Holoprosencephaly. European Journal of Human Genetics, 2011, 19, 3-3.	2.8	15
27	Regulation of downstream neuronal genes by proneural transcription factors during initial neurogenesis in the vertebrate brain. Neural Development, 2016, 11, 22.	2.4	15
28	Targeted panel sequencing establishes the implication of planar cell polarity pathway and involves new candidate genes in neural tube defect disorders. Human Genetics, 2019, 138, 363-374.	3.8	15
29	Disrupted Hypothalamo-Pituitary Axis in Association With Reduced SHH Underlies the Pathogenesis of NOTCH-Deficiency. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e3183-e3196.	3.6	10