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List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	Zebrafish xenografts as a tool for <i>in vivo</i> studies on human cancer. Annals of the New York Academy of Sciences, 2012, 1266, 124-137.	3.8	186
2	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 356-370.	6.2	171
3	BCL11A deletions result in fetal hemoglobin persistence and neurodevelopmental alterations. Journal of Clinical Investigation, 2015, 125, 2363-2368.	8.2	122
4	Debunking Occam's razor: Diagnosing multiple genetic diseases in families by wholeâ€exome sequencing. Clinical Genetics, 2017, 92, 281-289.	2.0	92
5	Clinical epigenomics: genome-wide DNA methylation analysis for the diagnosis of Mendelian disorders. Genetics in Medicine, 2021, 23, 1065-1074.	2.4	88
6	Genotype-phenotype correlations in individuals with pathogenic <i>RERE</i> variants. Human Mutation, 2018, 39, 666-675.	2.5	34
7	Broad spectrum of neuropsychiatric phenotypes associated with white matter disease in <i>PTEN</i> hamartoma tumor syndrome. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 101-109.	1.7	34
8	The zebrafish reveals dependence of the mast cell lineage on Notch signaling in vivo. Blood, 2012, 119, 3585-3594.	1.4	33
9	The unsolved rare genetic disease atlas? An analysis of the unexplained phenotypic descriptions in OMIM®. , 2018, 178, 458-463.		25
10	Brain malformations in a patient with deletion 2p16.1: A refinement ofÂthe phenotype to BCL11A. European Journal of Medical Genetics, 2015, 58, 351-354.	1.3	24
11	Novel findings and expansion of phenotype in a mosaic <scp>RASopathy</scp> caused by somatic <scp><i>KRAS</i></scp> variants. American Journal of Medical Genetics, Part A, 2021, 185, 2829-2845.	1.2	23
12	A transgenic zebrafish model expressing <i><scp>KIT</scp></i> â€ <scp>D</scp> 816 <scp>V</scp> recapitulates features of aggressive systemic mastocytosis. British Journal of Haematology, 2014, 167, 48-61.	2.5	18
13	BCL11B-related disorder in two canadian children: Expanding the clinical phenotype. European Journal of Medical Genetics, 2020, 63, 104007.	1.3	17
14	Tattonâ€Brownâ€Rahman syndrome: Six individuals with novel features. American Journal of Medical Genetics, Part A, 2020, 182, 673-680.	1.2	11
15	A Unique Role of GATA1s in Down Syndrome Acute Megakaryocytic Leukemia Biology and Therapy. PLoS ONE, 2011, 6, e27486.	2.5	11
16	Haploinsufficiency of PRR12 causes a spectrum of neurodevelopmental, eye, and multisystem abnormalities. Genetics in Medicine, 2021, 23, 1234-1245.	2.4	6
17	Detection of α-Thalassemia by Using Multiplex Ligation-Dependent Probe Amplification as an Additional Method for Rare Mutations in Southern Turkey. Indian Journal of Hematology and Blood Transfusion, 2016, 32, 454-459.	0.6	5
18	Genetic Testing in Children with Epilepsy: Report of a Single-Center Experience. Canadian Journal of Neurological Sciences, 2021, 48, 233-244.	0.5	4

#	Article	IF	CITATIONS
19	Diagnosis of Variant Klinefelter Syndrome in a 21-Year-Old Male Who Presented with Sparse Facial Hair. Annals of Dermatology, 2012, 24, 368.	0.9	2
20	Phenotate: crowdsourcing phenotype annotations as exercises in undergraduate classes. Genetics in Medicine, 2020, 22, 1391-1400.	2.4	2
21	Brain Abnormalities in Patients with Germline Variants in <i>H3F3</i> : Novel Imaging Findings and Neurologic Symptoms Beyond Somatic Variants and Brain Tumors. American Journal of Neuroradiology, 2022, 43, 1048-1053.	2.4	2