Sixto GarcÃ-a-MiÃ±aÃºr

List of Publications by Year
in descending order

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35 papers	854 citations	623734 14 h-index	526287 27 g-index
37 all docs	37 docs citations	37 times ranked	1946 citing authors

#	Article	IF	Citations
1	Management of growth failure and other endocrine aspects in patients with Noonan syndrome across Europe: A sub-analysis of a European clinical practice survey. European Journal of Medical Genetics, 2022, 65, 104404.	1.3	6
2	The natural history of adults with Rubinstein-Taybi syndrome: a families-reported experience. European Journal of Human Genetics, 2022, , .	2.8	4
3	Variability in Phelan-McDermid Syndrome in a Cohort of 210 Individuals. Frontiers in Genetics, 2022, 13, 652454.	2.3	27
4	Molecular and histologic insights on early onset cardiomyopathy in Danon disease females. Clinical Genetics, 2021, 99, 481-483.	2.0	3
5	Delineation of the clinical and radiological features of <scp>Stuveâ€“Wiedemann</scp> syndrome childhood survivors, four new cases and review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 856-865.	1.2	4
6	Expanding the Phenotypic Spectrum of PAX6 Mutations: From Congenital Cataracts to Nystagmus. Genes, 2021, 12, 707.	2.4	8
7	Further validation and psychometric properties of the Spanish adaptation of the Genetic Counseling Outcome Scale. Journal of Genetic Counseling, 2021, , .	1.6	0
8	Mosaic Variegated Aneuploidy syndrome 2 caused by biallelic variants in CEP57, two new cases and review of the phenotype. European Journal of Medical Genetics, 2021, 64, 104338.	1.3	5
9	Management of cardiac aspects in children with Noonan syndrome â€“ results from a European clinical practice survey among paediatric cardiologists. European Journal of Medical Genetics, 2021, 65, 104372.	1.3	11
10	European Medical Education Initiative on Noonan syndrome: A clinical practice survey assessing the diagnosis and clinical management of individuals with Noonan syndrome across Europe. European Journal of Medical Genetics, 2021, 65, 104371.	1.3	3
11	A clinical scoring system for congenital contractural arachnodactyly. Genetics in Medicine, 2020, 22, 124-131.	2.4	17
12	Further delineation of neuropsychiatric findings in Tatton-Brown-Rahman syndrome due to disease-causing variants in DNMT3A: seven new patients. European Journal of Human Genetics, 2020, 28, 469-479.	2.8	16
13	Pathogenic variants in <scp>KPTN</scp>, a rare cause of macrocephaly and intellectual disability. American Journal of Medical Genetics, Part A, 2020, 182, 2222-2225.	1.2	6
14	New mutations associated with Hirschsprung disease. Anales De PediatrÃa (English Edition), 2020, 93, 222-227.	0.2	1
15	MAGEL2â€related disorders: A study and case series. Clinical Genetics, 2019, 96, 493-505.	2.0	26
16	MRX93 syndrome (BRWD3 gene): five new patients with novel mutations. Clinical Genetics, 2019, 95, 726-731.	2.0	13
17	New insights into genetic variant spectrum and genotypeâ€“phenotype correlations of Rubinsteinâ€Taybi syndrome in 39 CREBBPâ€positive patients. Molecular Genetics & Genomic Medicine, 2019, 7, e972.	1.2	18
18	Mutation update for the SATB2 gene. Human Mutation, 2019, 40, 1013-1029.	2.5	38

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19	Variable expressivity of syndromic BMP4-related eye, brain, and digital anomalies: A review of the literature and description of three new cases. European Journal of Human Genetics, 2019, 27, 1379-1388.	2.8	8
20	The Brain-Lung-Thyroid syndrome (BLTS): A novel deletion in chromosome 14q13.2-q21.1 expands the phenotype to humoral immunodeficiency. European Journal of Medical Genetics, 2018, 61, 393-398.	1.3	10
21	Differential regulation of twoFLNAtranscripts explains some of the phenotypic heterogeneity in the loss-of-function filaminopathies. Human Mutation, 2018, 39, 103-113.	2.5	24
22	Translation and Crossâ€Cultural Adaptation with Preliminary Validation of GCOSâ€24 for Use in Spain. Journal of Genetic Counseling, 2018, 27, 732-743.	1.6	10
23	In-frame Variants in FLNA Proximal Rod 1 Domain Associate With a Predominant Cardiac Valvular Phenotype. Revista Espanola De Cardiologia (English Ed), 2018, 71, 545-552.	0.6	1
24	Variance of IQ is partially dependent on deletion type among 1,427 22q11.2 deletion syndrome subjects. American Journal of Medical Genetics, Part A, 2018, 176, 2172-2181.	1.2	33
25	Genotype and phenotype spectrum of NRAS germline variants. European Journal of Human Genetics, 2017, 25, 823-831.	2.8	36
26	FOXP1-related intellectual disability syndrome: a recognisable entity. Journal of Medical Genetics, 2017, 54, 613-623.	3.2	48
27	Pitfalls of trio-based exome sequencing: imprinted genes and parental mosaicismâ€”MAGEL2 as an example. Genetics in Medicine, 2017, 19, 1283-1285.	2.4	10
28	Costello Syndrome and Umbilical Ligament Rhabdomyosarcoma in Two Pediatric Patients: Case Reports and Review of the Literature. Case Reports in Genetics, 2017, 2017, 1-13.	0.2	6
29	Clinical and molecular analyses of Beckwithâ€“Wiedemann syndrome: Comparison between spontaneous conception and assisted reproduction techniques. American Journal of Medical Genetics, Part A, 2016, 170, 2740-2749.	1.2	30
30	Two novel POC1A mutations in the primordial dwarfism, SOFT syndrome: Clinical homogeneity but also unreported malformations. American Journal of Medical Genetics, Part A, 2016, 170, 210-216.	1.2	14
31	Two Cases of Autosomal Recessive Congenital Ichthyosis due to CYP4F22 Mutations: Expanding the Genotype of Selfâ€Healing Collodion Baby. Pediatric Dermatology, 2016, 33, e48-51.	0.9	13
32	Practical guidelines for managing adults with 22q11.2 deletion syndrome. Genetics in Medicine, 2015, 17, 599-609.	2.4	222
33	Expanding the mutation spectrum in 182 Spanish probands with craniosynostosis: identification and characterization of novel TCF12 variants. European Journal of Human Genetics, 2015, 23, 907-914.	2.8	53
34	New microdeletion and microduplication syndromes: a comprehensive review. Genetics and Molecular Biology, 2014, 37, 210-219.	1.3	84
35	A de novo non-sense mutation in ZBTB18 in a patient with features of the 1q43q44 microdeletion syndrome. European Journal of Human Genetics, 2014, 22, 844-846.	2.8	45