Francesca C Radio

List of Publications by Year
in descending order

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50	774	567281 15	610901 24
papers	citations	h-index	g-index

51	51	51	1803
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations in KCNK4 that Affect Gating Cause a Recognizable Neurodevelopmental Syndrome. American Journal of Human Genetics, 2018, 103, 621-630.	6.2	73
2	Aberrant Function of the C-Terminal Tail of HIST1H1E Accelerates Cellular Senescence and Causes Premature Aging. American Journal of Human Genetics, 2019, 105, 493-508.	6.2	48
3	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	6.2	48
4	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	6.2	48
5	A novel missense mutation in SLC40A1 results in resistance to hepcidin and confirms the existence of two ferroportinâ€associated iron overload diseases. British Journal of Haematology, 2009, 147, 379-385.	2.5	42
6	Association of hepcidin promoter c.-582 A>G variant and iron overload in thalassemia major. Haematologica, 2009, 94, 1293-1296.	3.5	29
7	Hereditary hemochromatosis type 1 phenotype modifiers in Italian patients. The controversial role of variants in HAMP, BMP2, FTL and SLC40A1 genes. Blood Cells, Molecules, and Diseases, 2015, 55, 71-75.	1.4	29
8	Expanding the clinical and molecular spectrum of PRMT7 mutations: 3 additional patients and review. Clinical Genetics, 2018, 93, 675-681.	2.0	28
9	NBAS pathogenic variants: Defining the associated clinical and facial phenotype and genotypeâ€“phenotype correlations. Human Mutation, 2019, 40, 721-728.	2.5	26
10	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. Genetics in Medicine, 2020, 22, 1338-1347.	2.4	25
11	A second cohort of CHD3 patients expands the molecular mechanisms known to cause Snijders Blok-Campeau syndrome. European Journal of Human Genetics, 2020, 28, 1422-1431.	2.8	25
12	TBX2 gene duplication associated with complex heart defect and skeletal malformations. American Journal of Medical Genetics, Part A, 2010, 152A, 2061-2066.	1.2	23
13	SPRED2 loss-of-function causes a recessive Noonan syndrome-like phenotype. American Journal of Human Genetics, 2021, 108, 2112-2129.	6.2	23
14	POGZâ€related epilepsy: Case report and review of the literature. American Journal of Medical Genetics, Part A, 2019, 179, 1631-1636.	1.2	19
15	TFR2-related hereditary hemochromatosis as a frequent cause of primary iron overload in patients from Central-Southern Italy. Blood Cells, Molecules, and Diseases, 2014, 52, 83-87.	1.4	18
16	Bi-allelic LoF NRROS Variants Impairing Active TGF-Î²1 Delivery Cause a Severe Infantile-Onset Neurodegenerative Condition with Intracranial Calcification. American Journal of Human Genetics, 2020, 106, 559-569.	6.2	18
17	Variants of SOS2 are a rare cause of Noonan syndrome with particular predisposition for lymphatic complications. European Journal of Human Genetics, 2021, 29, 51-60.	2.8	17
18	When to test fetuses for RASopathies? Proposition from a systematic analysis of 352 multicenter cases and a postnatal cohort. Genetics in Medicine, 2021, 23, 1116-1124.	2.4	17

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19	Small 4p16.3 deletions: Three additional patients and review of the literature. American Journal of Medical Genetics, Part A, 2018, 176, 2501-2508.	1.2	16
20	Biallelic loss-of-function variants in PLD1 cause congenital right-sided cardiac valve defects and neonatal cardiomyopathy. Journal of Clinical Investigation, 2021, 131, .	8.2	16
21	Epilepsy and BRAF Mutations: Phenotypes, Natural History and Genotype-Phenotype Correlations. Genes, 2021, 12, 1316.	2.4	13
22	Proliferative vasculopathy and hydranencephalyâ€“hydrocephaly syndrome or Fowler syndrome: Report of a family and insight into the disease's mechanism. Molecular Genetics & Genomic Medicine, 2018, 6, 446-451.	1.2	11
23	Bi-allelic variants in SPATA5L1 lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy, and hearing loss. American Journal of Human Genetics, 2021, 108, 2006-2016.	6.2	11
24	Incremental net benefit of whole genome sequencing for newborns and children with suspected genetic disorders: Systematic review and meta-analysis of cost-effectiveness evidence. Health Policy, 2022, 126, 337-345.	3.0	11
25	Biallelic TRNT1 variants in a child with B cell immunodeficiency, periodic fever and developmental delay without sideroblastic anemia (SIFD variant). Immunology Letters, 2020, 225, 64-65.	2.5	10
26	<scp>TSPEAR</scp> variants are primarily associated with ectodermal dysplasia and tooth agenesis but not hearing loss: A novel cohort study. American Journal of Medical Genetics, Part A, 2021, 185, 2417-2433.	1.2	10
27	Type 3 hereditary hemochromatosis in a patient from sub-Saharan Africa: Is there a link between African iron overload and TFR2 dysfunction?. Blood Cells, Molecules, and Diseases, 2013, 50, 31-32.	1.4	9
28	Functional analysis of TLK2 variants and their proximal interactomes implicates impaired kinase activity and chromatin maintenance defects in their pathogenesis. Journal of Medical Genetics, 2022, 59, 170-179.	3.2	9
29	Deficiency of MFSD7c results in microcephaly-associated vasculopathy in Fowler syndrome. Journal of Clinical Investigation, 2020, 130, 4081-4093.	8.2	9
30	<scp>PPP1R21</scp>â€related syndromic intellectual disability: Report of an adult patient and review. American Journal of Medical Genetics, Part A, 2020, 182, 3014-3022.	1.2	8
31	A Novel CCT5 Missense Variant Associated with Early Onset Motor Neuropathy. International Journal of Molecular Sciences, 2020, 21, 7631.	4.1	8
32	Cost-effectiveness of exome sequencing: an Italian pilot study on undiagnosed patients. New Genetics and Society, 2019, 38, 249-263.	1.2	7
33	Complex Presentation of Hao-Fountain Syndrome Solved by Exome Sequencing Highlighting Co-Occurring Genomic Variants. Genes, 2022, 13, 889.	2.4	7
34	Hyperferritinemia-cataract syndrome: Long-term ophthalmic observations in an Italian family. Ophthalmic Genetics, 2016, 37, 318-322.	1.2	6
35	Characterization of three novel pathogenic SLC40A1 mutations and genotype/phenotype correlations in 7 Italian families with type 4 hereditary hemochromatosis. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 464-470.	3.8	6
36	A novel germline mutation in <scp>CDK4</scp> codon 24 associated to familial melanoma. Clinical Genetics, 2018, 93, 934-935.	2.0	6

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37	Expanding the clinical phenotype of the ultraâ€rare <scp>Skrabanâ€Deardorff</scp> syndrome: Two novel individuals with <scp>WDR26</scp> lossâ€ofâ€function variants and a literature review. American Journal of Medical Genetics, Part A, 2021, 185, 1712-1720.	1.2	6
38	Biallelic variants in <scp>ZNF142</scp> lead to a syndromic neurodevelopmental disorder. Clinical Genetics, 2022, 102, 98-109.	2.0	6
39	Natural history of TFR2â€related hereditary hemochromatosis in a 47â€yrâ€old Italian patient. European Journal of Haematology, 2009, 83, 494-496.	2.2	5
40	Refinement of the clinical and mutational spectrum of <scp>UBE2A</scp> deficiency syndrome. Clinical Genetics, 2020, 98, 172-178.	2.0	5
41	Adducted Thumb and Peripheral Polyneuropathy: Diagnostic Supports in Suspecting Whiteâ€“Sutton Syndrome: Case Report and Review of the Literature. Genes, 2021, 12, 950.	2.4	5
42	Sprengel anomaly in deletion 22q11.2 (DiGeorge/Veloâ€“Cardioâ€“Facial) syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 661-664.	1.2	3
43	Pathogenic PTPN11 variants involving the polyâ€glutamine Gln ²⁵⁵ â€Gln ²⁵⁶ â€Gln ²⁵⁷ stretch highlight the relevance of helix B in SHP2's functional regulation. Human Mutation, 2020, 41, 1171-1182.	2.5	3
44	Broadening the phenotypic spectrum of Beta3GalT6 â€associated phenotypes. American Journal of Medical Genetics, Part A, 2021, 185, 3153-3160.	1.2	3
45	Co-Occurring Heterozygous CNOT3 and SMAD6 Truncating Variants: Unusual Presentation and Refinement of the IDDSADF Phenotype. Genes, 2021, 12, 1009.	2.4	3
46	Posterior Lissencephaly Associated with Subcortical Band Heterotopia Due to a Variation in the CEP85L Gene: A Case Report and Refining of the Phenotypic Spectrum. Genes, 2021, 12, 1208.	2.4	2
47	<scp>SHP2</scp>'s gainâ€ofâ€function in <scp>Werner</scp> syndrome causes childhood disease onset likely resulting from negative genetic interaction. Clinical Genetics, 2022, 102, 12-21.	2.0	2
48	The â€œold themeâ€•of variability versus transitory phenotypes in thanatophoric dysplasia type 1: Two 19â€weekâ€old fetuses with (â€œSan Diegoâ€•variant) and without ragged metaphyses due to the same FGFR3 mutation. American Journal of Medical Genetics, Part A, 2013, 161, 2675-2677.	1.2	1
49	Whole Exome Sequencing Is the Minimal Technological Approach in Probands Born to Consanguineous Couples. Genes, 2021, 12, 962.	2.4	0
50	Elucidating the clinical spectrum and molecular basis of HYAL2 deficiency. Genetics in Medicine, 2022, 24, 631-644.	2.4	0