

Francesca C Radio

List of Publications by Year in descending order

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Version: 2024-02-01

50
papers

774
citations

567281

15
h-index

610901

24
g-index

51
all docs

51
docs citations

51
times ranked

1803
citing authors

#	ARTICLE	IF	CITATIONS
1	Functional analysis of <i>TLK2</i> variants and their proximal interactomes implicates impaired kinase activity and chromatin maintenance defects in their pathogenesis. <i>Journal of Medical Genetics</i> , 2022, 59, 170-179.	3.2	9
2	Elucidating the clinical spectrum and molecular basis of <i>HYAL2</i> deficiency. <i>Genetics in Medicine</i> , 2022, 24, 631-644.	2.4	0
3	Incremental net benefit of whole genome sequencing for newborns and children with suspected genetic disorders: Systematic review and meta-analysis of cost-effectiveness evidence. <i>Health Policy</i> , 2022, 126, 337-345.	3.0	11
4	<i>SHP2</i> 's gain-of-function in <i>Werner</i> syndrome causes childhood disease onset likely resulting from negative genetic interaction. <i>Clinical Genetics</i> , 2022, 102, 12-21.	2.0	2
5	Complex Presentation of Hao-Fountain Syndrome Solved by Exome Sequencing Highlighting Co-Occurring Genomic Variants. <i>Genes</i> , 2022, 13, 889.	2.4	7
6	Biallelic variants in <i>ZNF142</i> lead to a syndromic neurodevelopmental disorder. <i>Clinical Genetics</i> , 2022, 102, 98-109.	2.0	6
7	Variants of <i>SOS2</i> are a rare cause of Noonan syndrome with particular predisposition for lymphatic complications. <i>European Journal of Human Genetics</i> , 2021, 29, 51-60.	2.8	17
8	When to test fetuses for RASopathies? Proposition from a systematic analysis of 352 multicenter cases and a postnatal cohort. <i>Genetics in Medicine</i> , 2021, 23, 1116-1124.	2.4	17
9	Expanding the clinical phenotype of the ultra-rare <i>Skraban-Deardorff</i> syndrome: Two novel individuals with <i>WDR26</i> loss-of-function variants and a literature review. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1712-1720.	1.2	6
10	<i>SPEN</i> haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an epismature of X chromosomes in females. <i>American Journal of Human Genetics</i> , 2021, 108, 502-516.	6.2	48
11	Biallelic loss-of-function variants in <i>PLD1</i> cause congenital right-sided cardiac valve defects and neonatal cardiomyopathy. <i>Journal of Clinical Investigation</i> , 2021, 131, .	8.2	16
12	<i>TSPEAR</i> variants are primarily associated with ectodermal dysplasia and tooth agenesis but not hearing loss: A novel cohort study. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2417-2433.	1.2	10
13	Broadening the phenotypic spectrum of <i>Beta3GalT6</i> associated phenotypes. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3153-3160.	1.2	3
14	Co-Occurring Heterozygous <i>CNOT3</i> and <i>SMAD6</i> Truncating Variants: Unusual Presentation and Refinement of the <i>IDDSADF</i> Phenotype. <i>Genes</i> , 2021, 12, 1009.	2.4	3
15	Adducted Thumb and Peripheral Polyneuropathy: Diagnostic Supports in Suspecting White-Sutton Syndrome: Case Report and Review of the Literature. <i>Genes</i> , 2021, 12, 950.	2.4	5
16	Whole Exome Sequencing Is the Minimal Technological Approach in Proband Born to Consanguineous Couples. <i>Genes</i> , 2021, 12, 962.	2.4	0
17	Posterior Lissencephaly Associated with Subcortical Band Heterotopia Due to a Variation in the <i>CEP85L</i> Gene: A Case Report and Refining of the Phenotypic Spectrum. <i>Genes</i> , 2021, 12, 1208.	2.4	2
18	Epilepsy and <i>BRAF</i> Mutations: Phenotypes, Natural History and Genotype-Phenotype Correlations. <i>Genes</i> , 2021, 12, 1316.	2.4	13

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19	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
20	SPRED2 loss-of-function causes a recessive Noonan syndrome-like phenotype. American Journal of Human Genetics, 2021, 108, 2112-2129.	6.2	23
21	<i>PPP1R21</i>-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
22	A Novel CCT5 Missense Variant Associated with Early Onset Motor Neuropathy. International Journal of Molecular Sciences, 2020, 21, 7631.	4.1	8
23	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	6.2	48
24	Refinement of the clinical and mutational spectrum of <sc>UBE2A</sc> deficiency syndrome. Clinical Genetics, 2020, 98, 172-178.	2.0	5
25	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
26	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
27	Bi-allelic LoF NRROS Variants Impairing Active TGF- β 1 Delivery Cause a Severe Infantile-Onset Neurodegenerative Condition with Intracranial Calcification. American Journal of Human Genetics, 2020, 106, 559-569.	6.2	18
28	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
29	Pathogenic <i>PTPN11</i> variants involving the poly-glutamine Gln ²⁵⁵ -Gln ²⁵⁶ -Gln ²⁵⁷ stretch highlight the relevance of helix B in SHP2's functional regulation. Human Mutation, 2020, 41, 1171-1182.	2.5	3
30	Deficiency of MFSD7c results in microcephaly-associated vasculopathy in Fowler syndrome. Journal of Clinical Investigation, 2020, 130, 4081-4093.	8.2	9
31	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
32	POGZ-related epilepsy: Case report and review of the literature. American Journal of Medical Genetics, Part A, 2019, 179, 1631-1636.	1.2	19
33	Cost-effectiveness of exome sequencing: an Italian pilot study on undiagnosed patients. New Genetics and Society, 2019, 38, 249-263.	1.2	7
34	<i>NBAS</i> pathogenic variants: Defining the associated clinical and facial phenotype and genotype-phenotype correlations. Human Mutation, 2019, 40, 721-728.	2.5	26
35	Proliferative vasculopathy and hydranencephaly-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
36	Expanding the clinical and molecular spectrum of <i>PRMT7</i> mutations: 3 additional patients and review. Clinical Genetics, 2018, 93, 675-681.	2.0	28

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37	Characterization of three novel pathogenic SLC40A1 mutations and genotype/phenotype correlations in 7 Italian families with type 4 hereditary hemochromatosis. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2018, 1864, 464-470.	3.8	6
38	A novel germline mutation in <i>CDK4</i> codon 24 associated to familial melanoma. <i>Clinical Genetics</i> , 2018, 93, 934-935.	2.0	6
39	Small 4p16.3 deletions: Three additional patients and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2501-2508.	1.2	16
40	Mutations in <i>KCNK4</i> that Affect Gating Cause a Recognizable Neurodevelopmental Syndrome. <i>American Journal of Human Genetics</i> , 2018, 103, 621-630.	6.2	73
41	Sprengel anomaly in deletion 22q11.2 (DiGeorge/Velo "Cardio" "Facial) syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 661-664.	1.2	3
42	Hyperferritinemia-cataract syndrome: Long-term ophthalmic observations in an Italian family. <i>Ophthalmic Genetics</i> , 2016, 37, 318-322.	1.2	6
43	Hereditary hemochromatosis type 1 phenotype modifiers in Italian patients. The controversial role of variants in <i>HAMP</i> , <i>BMP2</i> , <i>FTL</i> and <i>SLC40A1</i> genes. <i>Blood Cells, Molecules, and Diseases</i> , 2015, 55, 71-75.	1.4	29
44	TFR2-related hereditary hemochromatosis as a frequent cause of primary iron overload in patients from Central-Southern Italy. <i>Blood Cells, Molecules, and Diseases</i> , 2014, 52, 83-87.	1.4	18
45	The "old theme" of variability versus transitory phenotypes in thanatophoric dysplasia type 1: Two 19-week-old fetuses with ("San Diego" variant) and without ragged metaphyses due to the same <i>FGFR3</i> mutation. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2675-2677.	1.2	1
46	Type 3 hereditary hemochromatosis in a patient from sub-Saharan Africa: Is there a link between African iron overload and TFR2 dysfunction?. <i>Blood Cells, Molecules, and Diseases</i> , 2013, 50, 31-32.	1.4	9
47	<i>TBX2</i> gene duplication associated with complex heart defect and skeletal malformations. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 2061-2066.	1.2	23
48	Association of hepcidin promoter c.-582 A>G variant and iron overload in thalassemia major. <i>Haematologica</i> , 2009, 94, 1293-1296.	3.5	29
49	Natural history of TFR2-related hereditary hemochromatosis in a 47-year-old Italian patient. <i>European Journal of Haematology</i> , 2009, 83, 494-496.	2.2	5
50	A novel missense mutation in <i>SLC40A1</i> results in resistance to hepcidin and confirms the existence of two ferroportin-associated iron overload diseases. <i>British Journal of Haematology</i> , 2009, 147, 379-385.	2.5	42