

# Danilo Moretti-Ferreira

## List of Publications by Year in descending order

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43  
papers

1,449  
citations

623699

14  
h-index

330122

37  
g-index

44  
all docs

44  
docs citations

44  
times ranked

2019  
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in IRF6 cause Van der Woude and popliteal pterygium syndromes. <i>Nature Genetics</i> , 2002, 32, 285-289.	21.4	784
2	Prevalence and nonrandom distribution of exonic mutations in interferon regulatory factor 6 in 307 families with Van der Woude syndrome and 37 families with popliteal pterygium syndrome. <i>Genetics in Medicine</i> , 2009, 11, 241-247.	2.4	110
3	Williams-Beuren syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1128-1136.	1.2	55
4	Cornelia de Lange syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 150-158.	1.2	40
5	High Dosage Folic Acid Supplementation, Oral Cleft Recurrence and Fetal Growth. <i>International Journal of Environmental Research and Public Health</i> , 2013, 10, 590-605.	2.6	34
6	X-linked intellectual disability related genes disrupted by balanced X-autosome translocations. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2015, 168, 669-677.	1.7	32
7	Mucopolidosis types II and III and non-syndromic stuttering are associated with different variants in the same genes. <i>European Journal of Human Genetics</i> , 2016, 24, 529-534.	2.8	32
8	Macrocephaly, multiple lipomas, and hemangiomata (Bannayan-Zonana syndrome): Genetic heterogeneity or autosomal dominant locus with at least two different allelic forms?. <i>American Journal of Medical Genetics Part A</i> , 1989, 34, 548-551.	2.4	29
9	Macrosomia, obesity, macrocephaly and ocular abnormalities (MOMO syndrome) in two unrelated patients: Delineation of a newly recognized overgrowth syndrome. <i>American Journal of Medical Genetics Part A</i> , 1993, 46, 555-558.	2.4	28
10	Oral cleft prevention program (OCP). <i>BMC Pediatrics</i> , 2012, 12, 184.	1.7	28
11	X-chromosome inactivation patterns in monozygotic twins and sib pairs discordant for nonsyndromic cleft lip and/or palate. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 3267-3272.	1.2	20
12	Detection of classical 17p11.2 deletions, an atypical deletion and RAI1 alterations in patients with features suggestive of Smith-Magenis syndrome. <i>European Journal of Human Genetics</i> , 2012, 20, 148-154.	2.8	18
13	Incorporation of 5-ethynyl-2-deoxyuridine (EdU) as a novel strategy for identification of the skewed X inactivation pattern in balanced and unbalanced X-rearrangements. <i>Human Genetics</i> , 2016, 135, 185-192.	3.8	18
14	Search for Genomic Alterations in Monozygotic Twins Discordant for Cleft Lip and/or Palate. <i>Twin Research and Human Genetics</i> , 2009, 12, 462-468.	0.6	16
15	A genetic linkage study in Brazil identifies a new locus for persistent developmental stuttering on chromosome 10. <i>Genetics and Molecular Research</i> , 2014, 13, 2094-2101.	0.2	16
16	Rubinstein-Taybi syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2939-2950.	1.2	16
17	A study of the role of the FOXP2 and CNTNAP2 genes in persistent developmental stuttering. <i>Neurobiology of Disease</i> , 2014, 69, 23-31.	4.4	15
18	Sulcus vocalis: evidence for autosomal dominant inheritance. <i>Genetics and Molecular Research</i> , 2011, 10, 3163-3168.	0.2	14

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19	Evaluation of the association between polymorphisms at the DRD2 locus and stuttering. <i>Journal of Human Genetics</i> , 2011, 56, 472-473.	2.3	14
20	Perfil da fluência da fala na síndrome de Williams-Beuren: estudo preliminar. <i>Prá-fono: Revista De Atualizaçáo Científica</i> , 2009, 21, 107-112.	0.5	13
21	Genomic strategy identifies a missense mutation in <i>WD-repeat domain 65</i> ( <i>WDR65</i> ) in an individual with Van der Woude syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1314-1321.	1.2	12
22	Cytogenetic Evidence of Involvement of Chromosome Regions 15q12 and 12q15 in Conditions with Associated Overgrowth. <i>DNA and Cell Biology</i> , 1993, 12, 227-231.	1.9	11
23	Fluorescent in situ hybridization (FISH) as a diagnostic tool for Williams-Beuren syndrome. <i>Genetics and Molecular Biology</i> , 2007, 30, 17-20.	1.3	9
24	X monosomy and balanced Robertsonian translocation in a girl with Turner Syndrome. <i>Genetics and Molecular Biology</i> , 2006, 29, 47-48.	1.3	9
25	DHCR7 mutations in Brazilian Smith-Lemli-Opitz syndrome patients. <i>American Journal of Medical Genetics, Part A</i> , 2005, 136A, 278-281.	1.2	8
26	Differential diagnosis of Smith-Magenis syndrome: 1p36 deletion syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 988-992.	1.2	7
27	Metacarpophalangeal pattern profile in Marfan syndrome and Marfan-like patients. , 1997, 72, 159-163.		6
28	Folic Acid Fortification and Women's Folate Levels in Selected Communities in Brazil - A First Look. <i>International Journal for Vitamin and Nutrition Research</i> , 2014, 84, 286-294.	1.5	6
29	MOMO syndrome associated with autism: a case report. <i>Genetics and Molecular Research</i> , 2008, 7, 1223-1225.	0.2	6
30	Transmission analysis of candidate genes for nonsyndromic oral clefts in Brazilian parent-child triads with recurrence. <i>Genetics and Molecular Biology</i> , 2006, 29, 439-442.	1.3	5
31	Short Communication Assessment of clinical scoring systems for the diagnosis of Williams-Beuren syndrome. <i>Genetics and Molecular Research</i> , 2013, 12, 3407-3411.	0.2	5
32	Autism Spectrum Disorder in a Girl with a <i>De Novo</i> X;19 Balanced Translocation. <i>Case Reports in Genetics</i> , 2012, 2012, 1-4.	0.2	4
33	Are variants in sex hormone metabolizing genes associated with stuttering?. <i>Brain and Language</i> , 2019, 191, 28-30.	1.6	4
34	Case Report Oropharyngeal dysphagia and language delay in partial trisomy 9p: case report. <i>Genetics and Molecular Research</i> , 2009, 8, 1133-1138.	0.2	4
35	Smith-Lemli-Opitz syndrome: clinical and biochemical findings in Brazilian patients. <i>Genetics and Molecular Biology</i> , 2006, 29, 429-436.	1.3	3
36	Schilbach-Rott/blepharofacioskeletal syndrome in a Brazilian patient. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 2134-2137.	1.2	3

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37	Clinical and Molecular Heterogeneity in Brazilian Patients with Sotos Syndrome. <i>Molecular Syndromology</i> , 2015, 6, 32-38.	0.8	3
38	Diagnosis of Smith-Lemli-Opitz syndrome by ultraviolet spectrophotometry. <i>Brazilian Journal of Medical and Biological Research</i> , 2003, 36, 1327-1332.	1.5	2
39	Spread of X chromosome inactivation into autosomal regions in patients with unbalanced X-autosome translocations and its phenotypic effects. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2295-2305.	1.2	2
40	A rare non-Robertsonian translocation involving chromosomes 15 and 21. <i>Sao Paulo Medical Journal</i> , 2013, 131, 427-431.	0.9	1
41	Schilbach-Rott syndrome associated with 9q22.32q22.33 duplication, involving the PTCH1 gene. <i>European Journal of Human Genetics</i> , 2019, 27, 1260-1266.	2.8	1
42	Cover Image, Volume 176A, Number 5, May 2018. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, .	1.2	0
43	Non-mosaic partial duplication 12p in a patient with dysmorphic characteristics and developmental delay. <i>Genetics and Molecular Biology</i> , 2020, 43, e20180285.	1.3	0