

Doriana Misceo

List of Publications by Year in descending order

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Version: 2024-02-01

85
papers

2,564
citations

257450

24
h-index

223800

46
g-index

86
all docs

86
docs citations

86
times ranked

4037
citing authors

#	ARTICLE	IF	CITATIONS
1	DNA methylation epsignature in Gabriele-de VriesÂsyndrome. <i>Genetics in Medicine</i> , 2022, 24, 905-914.	2.4	6
2	ZBTB11 dysfunction: spectrum of brain abnormalities, biochemical signature and cellular consequences. <i>Brain</i> , 2022, 145, 2602-2616.	7.6	5
3	STIM1 R304W in mice causes subgingival hair growth and an increased fraction of trabecular bone. <i>Cell Calcium</i> , 2020, 85, 102110.	2.4	8
4	The expanding clinical phenotype of germline <i>ABL1</i> associated congenital heart defects and skeletal malformations syndrome. <i>Human Mutation</i> , 2020, 41, 1738-1744.	2.5	10
5	Loss of CBY1 results in a ciliopathy characterized by features of Joubert syndrome. <i>Human Mutation</i> , 2020, 41, 2179-2194.	2.5	16
6	Sudden death in epilepsy and ectopic neurohypophysis in Joubert syndrome 23 diagnosed using SNVs/indels and structural variants pipelines on WGS data: a case report. <i>BMC Medical Genetics</i> , 2020, 21, 96.	2.1	5
7	Biallelic variants in the RNA exosome gene <i>EXOSC5</i> are associated with developmental delays, short stature, cerebellar hypoplasia and motor weakness. <i>Human Molecular Genetics</i> , 2020, 29, 2218-2239.	2.9	19
8	A girl with a neurodevelopmental syndrome, adducted thumbs and frequent infections caused by novel homozygous variant in <i>DEAF1</i> . <i>Clinical Dysmorphology</i> , 2020, 29, 107-110.	0.3	2
9	Closed-Form Saint-Venant Solutions in the Koiter Theory of Shells. <i>Journal of Elasticity</i> , 2020, 140, 149-169.	1.9	1
10	Refined dimensional reduction for isotropic elastic Cosserat shells with initial curvature. <i>Mathematics and Mechanics of Solids</i> , 2019, 24, 4000-4019.	2.4	15
11	TBCK Encephaloneuropathy With Abnormal Lysosomal Storage: Use of a Structural Variant Bioinformatics Pipeline on Whole-Genome Sequencing Data Unravels a 20-Year-Old Clinical Mystery. <i>Pediatric Neurology</i> , 2019, 96, 74-75.	2.1	11
12	Human TGFÎ²1 deficiency causes severe inflammatory bowel disease and encephalopathy. <i>Nature Genetics</i> , 2018, 50, 344-348.	21.4	95
13	Biallelic variants in <i>LINGO1</i> are associated with autosomal recessive intellectual disability, microcephaly, speech and motor delay. <i>Genetics in Medicine</i> , 2018, 20, 778-784.	2.4	21
14	Mutated Thyroid Hormone Transporter <i>OATP1C1</i> Associates with Severe Brain Hypometabolism and Juvenile Neurodegeneration. <i>Thyroid</i> , 2018, 28, 1406-1415.	4.5	57
15	STIM1 R304W causes muscle degeneration and impaired platelet activation in mice. <i>Cell Calcium</i> , 2018, 76, 87-100.	2.4	21
16	YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. <i>American Journal of Human Genetics</i> , 2017, 100, 907-925.	6.2	125
17	Segregation of Incomplete Achromatopsia and Alopecia Due to <i>PDE6H</i> and <i>LPAR6</i> Variants in a Consanguineous Family from Pakistan. <i>Genes</i> , 2016, 7, 41.	2.4	8
18	Novel <i>PIGT</i> Variant in Two Brothers: Expansion of the Multiple Congenital Anomalies-Hypotonia Seizures Syndrome 3 Phenotype. <i>Genes</i> , 2016, 7, 108.	2.4	25

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19	A novel mutation in FBXL4 in a Norwegian child with encephalomyopathic mitochondrial DNA depletion syndrome 13. <i>European Journal of Medical Genetics</i> , 2016, 59, 342-346.	1.3	16
20	Numerical treatment of a geometrically nonlinear planar Cosserat shell model. <i>Computational Mechanics</i> , 2016, 57, 817-841.	4.0	27
21	Delineating the <i>GRIN1</i> phenotypic spectrum. <i>Neurology</i> , 2016, 86, 2171-2178.	1.1	157
22	Clinical and molecular characteristics in three families with biallelic mutations in IGHMBP2. <i>Neuromuscular Disorders</i> , 2016, 26, 570-575.	0.6	17
23	Haploinsufficiency of <i>ANO6</i> , <i>NELL2</i> and <i>DBX2</i> in a boy with intellectual disability and growth delay. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1890-1896.	1.2	13
24	A recurrent deletion on chromosome 2q13 is associated with developmental delay and mild facial dysmorphisms. <i>Molecular Cytogenetics</i> , 2015, 8, 57.	0.9	24
25	Kaufman oculocerebrofacial syndrome in sisters with novel compound heterozygous mutation in <i>UBE3B</i> . <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 657-663.	1.2	15
26	A novel type of rhizomelic chondrodysplasia punctata, RCDP5, is caused by loss of the PEX5 long isoform. <i>Human Molecular Genetics</i> , 2015, 24, 5845-5854.	2.9	73
27	Existence Theorem for Geometrically Nonlinear Cosserat Micropolar Model Under Uniform Convexity Requirements. <i>Journal of Elasticity</i> , 2015, 121, 119-141.	1.9	30
28	Multilayered and FGM structural elements under mechanical and thermal loads. Part I: Comparison of finite elements and analytical models. <i>Archives of Civil and Mechanical Engineering</i> , 2015, 15, 1180-1192.	3.8	21
29	Spastic Paraplegia Type 7 Is Associated with Multiple Mitochondrial DNA Deletions. <i>PLoS ONE</i> , 2014, 9, e86340.	2.5	49
30	Existence of minimizers in the geometrically non-linear 6-parameter resultant shell theory with drilling rotations. <i>Mathematics and Mechanics of Solids</i> , 2014, 19, 376-397.	2.4	39
31	Post-zygotic breakage of a dicentric chromosome results in mosaicism for a telocentric 9p marker chromosome in a boy with developmental delay. <i>Gene</i> , 2014, 533, 403-410.	2.2	10
32	A Dominant STIM1 Mutation Causes Stormorken Syndrome. <i>Human Mutation</i> , 2014, 35, 556-564.	2.5	143
33	A de novo 2.3Mb deletion in 2q24.2q24.3 in a 20-month-old developmentally delayed girl. <i>Gene</i> , 2014, 539, 168-172.	2.2	22
34	Shells without drilling rotations: A representation theorem in the framework of the geometrically nonlinear 6-parameter resultant shell theory. <i>International Journal of Engineering Science</i> , 2014, 80, 32-42.	5.0	16
35	Haploinsufficiency of XPO1 and USP34 by a de novo 230Åkb deletion in 2p15, in a patient with mild intellectual disability and cranio-facial dysmorphisms. <i>European Journal of Medical Genetics</i> , 2014, 57, 513-519.	1.3	18
36	Haploinsufficiency of two histone modifier genes on 6p22.3, ATXN1 and JARID2, is associated with intellectual disability. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 3.	2.7	18

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37	Sum of squared logarithms - an inequality relating positive definite matrices and their matrix logarithm. <i>Journal of Inequalities and Applications</i> , 2013, 2013, 168.	1.1	17
38	Mechanical behavior of sandwich composite beams made of foams and functionally graded materials. <i>International Journal of Solids and Structures</i> , 2013, 50, 519-530.	2.7	66
39	A balanced de novo inv(7)(p14.3q22.3) disrupting PDE1C and ATXN7L1 in a 14-year old developmentally delayed boy. <i>European Journal of Medical Genetics</i> , 2013, 56, 361-364.	1.3	3
40	Existence Theorems in the Geometrically Non-linear 6-Parameter Theory of Elastic Plates. <i>Journal of Elasticity</i> , 2013, 112, 185-198.	1.9	23
41	Thermoelastic Deformations of Cylindrical Multi-Layered Shells Using a Direct Approach. <i>Journal of Thermal Stresses</i> , 2013, 36, 749-789.	2.0	8
42	Hyperphagia, Mild Developmental Delay But Apparently No Structural Brain Anomalies in a Boy Without <i>SOX3</i> Expression. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1137-1142.	1.2	10
43	On the Cosserat model for thin rods made of thermoelastic materials with voids. <i>Discrete and Continuous Dynamical Systems - Series S</i> , 2013, 6, 1473-1485.	1.1	6
44	A mild form of Mucopolysaccharidosis IIIB diagnosed with targeted next-generation sequencing of linked genomic regions. <i>European Journal of Human Genetics</i> , 2012, 20, 58-63.	2.8	24
45	A 1.5Mb de novo deletion within 11q13.1q13.2 in a boy with mild intellectual disability and minor dysmorphic features. <i>European Journal of Medical Genetics</i> , 2012, 55, 695-699.	1.3	17
46	1.5Mb deletion of chromosome 4p16.3 associated with postnatal growth delay, psychomotor impairment, epilepsy, impulsive behavior and asynchronous skeletal development. <i>Gene</i> , 2012, 507, 85-91.	2.2	21
47	On a thermodynamic theory of rods with two temperature fields. <i>Acta Mechanica</i> , 2012, 223, 1583-1596.	2.1	37
48	Deformation analysis of functionally graded beams by the direct approach. <i>Composites Part B: Engineering</i> , 2012, 43, 1315-1328.	12.0	123
49	The Korn-type inequality in a Cosserat model for thin thermoelastic porous rods. <i>Meccanica</i> , 2012, 47, 789-794.	2.0	5
50	On a Problem of Truesdell for Anisotropic Elastic Shells. <i>Analele Stiintifice Ale Universitatii Al I Cuza Din Iasi - Matematica</i> , 2011, 57, .	0.2	0
51	Theory of thin thermoelastic rods made of porous materials. <i>Archive of Applied Mechanics</i> , 2011, 81, 1365-1391.	2.2	19
52	On the dynamical theory of thermoelastic simple shells. <i>ZAMM Zeitschrift Fur Angewandte Mathematik Und Mechanik</i> , 2011, 91, 443-457.	1.6	12
53	On the theory of porous elastic rods. <i>International Journal of Solids and Structures</i> , 2011, 48, 910-924.	2.7	25
54	Thermal stresses in anisotropic cylindrical elastic shells. <i>Mathematical Methods in the Applied Sciences</i> , 2010, 33, 799-810.	2.3	3

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55	A mathematical study of the linear theory for orthotropic elastic simple shells. <i>Mathematical Methods in the Applied Sciences</i> , 2010, 33, 1399-1413.	2.3	15
56	A de novo 15q13.2q13.3 deletion in a boy with an Angelman syndrome like phenotype. <i>European Journal of Medical Genetics</i> , 2010, 53, 221-224.	1.3	1
57	A partial trisomy 1q patient with a deletion 1q22 and an insertion 1(q42q44) into 1q22. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 290-293.	1.2	1
58	Inheritance of a terminal 7.1â€‰Mb 18p deletion flanked by a 2.3â€‰Mb duplication from a physically normal mother. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 2877-2881.	1.2	7
59	A satellite-like sequence, representing a "clone gap" in the human genome, was likely involved in the seeding of a novel centromere in macaque. <i>Chromosoma</i> , 2009, 118, 269-277.	2.2	9
60	On Saint-Venant's problem for anisotropic, inhomogeneous, cylindrical Cosserat elastic shells. <i>International Journal of Engineering Science</i> , 2009, 47, 21-38.	5.0	14
61	Thermal stresses in cylindrical Cosserat elastic shells. <i>European Journal of Mechanics, A/Solids</i> , 2009, 28, 94-101.	3.7	29
62	Refinement of macaque synteny arrangement with respect to the official rhesus macaque sequence assembly. <i>Chromosome Research</i> , 2008, 16, 977-985.	2.2	21
63	Inequalities of Korn's Type and Existence Results in the Theory of Cosserat Elastic Shells. <i>Journal of Elasticity</i> , 2008, 90, 227-239.	1.9	17
64	On the dynamic deformation of porous Cosserat linear thermoelastic shells. <i>ZAMM Zeitschrift Fur Angewandte Mathematik Und Mechanik</i> , 2008, 88, 74-78.	1.6	2
65	A de novo 6p interstitial deletion and a complex translocation involving chromosomes 2, 6, and 14 in a mildly developmentally delayed patient. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 3230-3233.	1.2	4
66	Tracking the complex flow of chromosome rearrangements from the Hominoidea Ancestor to extant <i>Hylobates</i> and <i>Nomascus</i> Gibbons by high-resolution synteny mapping. <i>Genome Research</i> , 2008, 18, 1530-1537.	5.5	41
67	On the Bending Equations for Elastic Plates with Voids. <i>Mathematics and Mechanics of Solids</i> , 2007, 12, 40-57.	2.4	2
68	Evolutionary history of chromosome 11 featuring four distinct centromere repositioning events in <i>Catarrhini</i> . <i>Genomics</i> , 2007, 90, 35-43.	2.9	28
69	On the theory of loaded general cylindrical Cosserat elastic shells. <i>International Journal of Solids and Structures</i> , 2007, 44, 7399-7419.	2.7	6
70	On Saint-Venant's principle in the theory of Cosserat elastic shells. <i>International Journal of Engineering Science</i> , 2007, 45, 187-198.	5.0	19
71	On a Thermodynamic Theory of Porous Cosserat Elastic Shells. <i>Journal of Thermal Stresses</i> , 2006, 29, 879-899.	2.0	20
72	Evolutionary movement of centromeres in horse, donkey, and zebra. <i>Genomics</i> , 2006, 87, 777-782.	2.9	100

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73	On the theory of elastic shells made from a material with voids. <i>International Journal of Solids and Structures</i> , 2006, 43, 3106-3123.	2.7	18
74	Several results in the dynamic theory of thermoelastic Cosserat shells with voids. <i>Mechanics Research Communications</i> , 2006, 33, 157-176.	1.8	14
75	A preliminary comparative analysis of primate segmental duplications shows elevated substitution rates and a great-ape expansion of intrachromosomal duplications. <i>Genome Research</i> , 2006, 16, 576-583.	5.5	82
76	Saint-Venant's problem for Cosserat shells with voids. <i>International Journal of Solids and Structures</i> , 2005, 42, 2033-2057.	2.7	17
77	Minimum Energy Characterizations for the Solution of Saint-Venant's Problem in the Theory of Shells. <i>Journal of Elasticity</i> , 2005, 81, 179-204.	1.9	4
78	Recurrent Sites for New Centromere Seeding. <i>Genome Research</i> , 2004, 14, 1696-1703.	5.5	135
79	Evolutionary History of Chromosome 20. <i>Molecular Biology and Evolution</i> , 2004, 22, 360-366.	8.9	21
80	The Solution of Saint-Venant's Problem in the Theory of Cosserat Shells. <i>Journal of Elasticity</i> , 2004, 74, 185-214.	1.9	24
81	Human chromosome 16 conservation in primates. <i>Chromosome Research</i> , 2003, 11, 323-326.	2.2	12
82	Refinement of a chimpanzee pericentric inversion breakpoint to a segmental duplication cluster. <i>Genome Biology</i> , 2003, 4, R50.	9.6	107
83	A BENDING THEORY OF POROUS THERMOELASTIC PLATES. <i>Journal of Thermal Stresses</i> , 2003, 26, 67-90.	2.0	25
84	Human Paralogs of <i>KIAA0187</i> Were Created through Independent Pericentromeric-Directed and Chromosome-Specific Duplication Mechanisms. <i>Genome Research</i> , 2002, 12, 67-80.	5.5	24
85	Human-Specific Duplication and Mosaic Transcripts: The Recent Paralogous Structure of Chromosome 22. <i>American Journal of Human Genetics</i> , 2002, 70, 83-100.	6.2	168