Doriana Misceo

List of Publications by Year
in descending order

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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	Human-Specific Duplication and Mosaic Transcripts: The Recent Paralogous Structure of Chromosome 22. American Journal of Human Genetics, 2002, 70, 83-100.	6.2	168
2	Delineating the GRIN1 phenotypic spectrum. Neurology, 2016, 86, 2171-2178.	1.1	157
3	A Dominant STIM1 Mutation Causes Stormorken Syndrome. Human Mutation, 2014, 35, 556-564.	2.5	143
4	Recurrent Sites for New Centromere Seeding. Genome Research, 2004, 14, 1696-1703.	5.5	135
5	YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. American Journal of Human Genetics, 2017, 100, 907-925.	6.2	125
6	Deformation analysis of functionally graded beams by the direct approach. Composites Part B: Engineering, 2012, 43, 1315-1328.	12.0	123
7	Refinement of a chimpanzee pericentric inversion breakpoint to a segmental duplication cluster. Genome Biology, 2003, 4, R50.	9.6	107
8	Evolutionary movement of centromeres in horse, donkey, and zebra. Genomics, 2006, 87, 777-782.	2.9	100
9	Human TGF-Î²1 deficiency causes severe inflammatory bowel disease and encephalopathy. Nature Genetics, 2018, 50, 344-348.	21.4	95
10	A preliminary comparative analysis of primate segmental duplications shows elevated substitution rates and a great-ape expansion of intrachromosomal duplications. Genome Research, 2006, 16, 576-583.	5.5	82
11	A novel type of rhizomelic chondrodysplasia punctata, RCDP5, is caused by loss of the PEX5 long isoform. Human Molecular Genetics, 2015, 24, 5845-5854.	2.9	73
12	Mechanical behavior of sandwich composite beams made of foams and functionally graded materials. International Journal of Solids and Structures, 2013, 50, 519-530.	2.7	66
13	Mutated Thyroid Hormone Transporter OATP1C1 Associates with Severe Brain Hypometabolism and Juvenile Neurodegeneration. Thyroid, 2018, 28, 1406-1415.	4.5	57
14	Spastic Paraplegia Type 7 Is Associated with Multiple Mitochondrial DNA Deletions. PLoS ONE, 2014, 9, e86340.	2.5	49
15	Tracking the complex flow of chromosome rearrangements from the Hominoidea Ancestor to extant Hylobates and Nomascus Gibbons by high-resolution synteny mapping. Genome Research, 2008, 18, 1530-1537.	5.5	41
16	Existence of minimizers in the geometrically non-linear 6-parameter resultant shell theory with drilling rotations. Mathematics and Mechanics of Solids, 2014, 19, 376-397.	2.4	39
17	On a thermodynamic theory of rods with two temperature fields. Acta Mechanica, 2012, 223, 1583-1596.	2.1	37
18	Existence Theorem for Geometrically Nonlinear Cosserat Micropolar Model Under Uniform Convexity Requirements. Journal of Elasticity, 2015, 121, 119-141.	1.9	30

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19	Thermal stresses in cylindrical Cosserat elastic shells. European Journal of Mechanics, A/Solids, 2009, 28, 94-101.	3.7	29
20	Evolutionary history of chromosome 11 featuring four distinct centromere repositioning events in Catarrhini. Genomics, 2007, 90, 35-43.	2.9	28
21	Numerical treatment of a geometrically nonlinear planar Cosserat shell model. Computational Mechanics, 2016, 57, 817-841.	4.0	27
22	A BENDING THEORY OF POROUS THERMOELASTIC PLATES. Journal of Thermal Stresses, 2003, 26, 67-90.	2.0	25
23	On the theory of porous elastic rods. International Journal of Solids and Structures, 2011, 48, 910-924.	2.7	25
24	Novel PIGT Variant in Two Brothers: Expansion of the Multiple Congenital Anomalies-Hypotonia Seizures Syndrome 3 Phenotype. Genes, 2016, 7, 108.	2.4	25
25	Human Paralogs of KIAA0187 Were Created through Independent Pericentromeric-Directed and Chromosome-Specific Duplication Mechanisms. Genome Research, 2002, 12, 67-80.	5.5	24
26	The Solution of Saint-Venant's Problem in the Theory of Cosserat Shells. Journal of Elasticity, 2004, 74, 185-214.	1.9	24
27	A mild form of Mucopolysaccharidosis IIIB diagnosed with targeted next-generation sequencing of linked genomic regions. European Journal of Human Genetics, 2012, 20, 58-63.	2.8	24
28	A recurrent deletion on chromosome 2q13 is associated with developmental delay and mild facial dysmorphisms. Molecular Cytogenetics, 2015, 8, 57.	0.9	24
29	Existence Theorems in the Geometrically Non-linear 6-Parameter Theory of Elastic Plates. Journal of Elasticity, 2013, 112, 185-198.	1.9	23
30	A de novo 2.3Mb deletion in 2q24.2q24.3 in a 20-month-old developmentally delayed girl. Gene, 2014, 539, 168-172.	2.2	22
31	Evolutionary History of Chromosome 20. Molecular Biology and Evolution, 2004, 22, 360-366.	8.9	21
32	Refinement of macaque synteny arrangement with respect to the official rheMac2 macaque sequence assembly. Chromosome Research, 2008, 16, 977-985.	2.2	21
33	1.5Mb deletion of chromosome 4p16.3 associated with postnatal growth delay, psychomotor impairment, epilepsy, impulsive behavior and asynchronous skeletal development. Gene, 2012, 507, 85-91.	2.2	21
34	Multilayered and FGM structural elements under mechanical and thermal loads. Part I: Comparison of finite elements and analytical models. Archives of Civil and Mechanical Engineering, 2015, 15, 1180-1192.	3.8	21
35	Biallelic variants in LINGO1 are associated with autosomal recessive intellectual disability, microcephaly, speech and motor delay. Genetics in Medicine, 2018, 20, 778-784.	2.4	21
36	STIM1 R304W causes muscle degeneration and impaired platelet activation in mice. Cell Calcium, 2018, 76, 87-100.	2.4	21

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37	On a Thermodynamic Theory of Porous Cosserat Elastic Shells. Journal of Thermal Stresses, 2006, 29, 879-899.	2.0	20
38	On Saint-Venantâ€™s principle in the theory of Cosserat elastic shells. International Journal of Engineering Science, 2007, 45, 187-198.	5.0	19
39	Theory of thin thermoelastic rods made of porous materials. Archive of Applied Mechanics, 2011, 81, 1365-1391.	2.2	19
40	Biallelic variants in the RNA exosome gene EXOSC5 are associated with developmental delays, short stature, cerebellar hypoplasia and motor weakness. Human Molecular Genetics, 2020, 29, 2218-2239.	2.9	19
41	On the theory of elastic shells made from a material with voids. International Journal of Solids and Structures, 2006, 43, 3106-3123.	2.7	18
42	Haploinsufficiency of two histone modifier genes on 6p22.3, ATXN1 and JARID2, is associated with intellectual disability. Orphanet Journal of Rare Diseases, 2013, 8, 3.	2.7	18
43	Haploinsufficiency of XPO1 and USP34 by a de novo 230Âkb deletion in 2p15, in a patient with mild intellectual disability and cranio-facial dysmorphisms. European Journal of Medical Genetics, 2014, 57, 513-519.	1.3	18
44	Saint-Venantâ€™s problem for Cosserat shells with voids. International Journal of Solids and Structures, 2005, 42, 2033-2057.	2.7	17
45	Inequalities of Kornâ€™s Type and Existence Results in the Theory of Cosserat Elastic Shells. Journal of Elasticity, 2008, 90, 227-239.	1.9	17
46	A 1ÂMb de novo deletion within 11q13.1q13.2 in a boy with mild intellectual disability and minor dysmorphic features. European Journal of Medical Genetics, 2012, 55, 695-699.	1.3	17
47	Sum of squared logarithms - an inequality relating positive definite matrices and their matrix logarithm. Journal of Inequalities and Applications, 2013, 2013, 168.	1.1	17
48	Clinical and molecular characteristics in three families with biallelic mutations in IGHMBP2. Neuromuscular Disorders, 2016, 26, 570-575.	0.6	17
49	Shells without drilling rotations: A representation theorem in the framework of the geometrically nonlinear 6-parameter resultant shell theory. International Journal of Engineering Science, 2014, 80, 32-42.	5.0	16
50	A novel mutation in FBXL4 in a Norwegian child with encephalomyopathic mitochondrial DNA depletion syndrome 13. European Journal of Medical Genetics, 2016, 59, 342-346.	1.3	16
51	Loss of CBY1 results in a ciliopathy characterized by features of Joubert syndrome. Human Mutation, 2020, 41, 2179-2194.	2.5	16
52	A mathematical study of the linear theory for orthotropic elastic simple shells. Mathematical Methods in the Applied Sciences, 2010, 33, 1399-1413.	2.3	15
53	Kaufman oculocerebrofacial syndrome in sisters with novel compound heterozygous mutation in UBE3B. American Journal of Medical Genetics, Part A, 2015, 167, 657-663.	1.2	15
54	Refined dimensional reduction for isotropic elastic Cosserat shells with initial curvature. Mathematics and Mechanics of Solids, 2019, 24, 4000-4019.	2.4	15

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55	Several results in the dynamic theory of thermoelastic Cosserat shells with voids. Mechanics Research Communications, 2006, 33, 157-176.	1.8	14
56	On Saintâ€“Venantâ€™s problem for anisotropic, inhomogeneous, cylindrical Cosserat elastic shells. International Journal of Engineering Science, 2009, 47, 21-38.	5.0	14
57	Haploinsufficiency of ANO6, NELL2 and DBX2 in a boy with intellectual disability and growth delay. American Journal of Medical Genetics, Part A, 2015, 167, 1890-1896.	1.2	13
58	Human chromosome 16 conservation in primates. Chromosome Research, 2003, 11, 323-326.	2.2	12
59	On the dynamical theory of thermoelastic simple shells. ZAMM Zeitschrift Fur Angewandte Mathematik Und Mechanik, 2011, 91, 443-457.	1.6	12
60	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. Pediatric Neurology, 2019, 96, 74-75.	2.1	11
61	Hyperphagia, Mild Developmental Delay But Apparently No Structural Brain Anomalies in a Boy Without <scp>SOX</scp>3 Expression. American Journal of Medical Genetics, Part A, 2013, 161, 1137-1142.	1.2	10
62	Post-zygotic breakage of a dicentric chromosome results in mosaicism for a telocentric 9p marker chromosome in a boy with developmental delay. Gene, 2014, 533, 403-410.	2.2	10
63	The expanding clinical phenotype of germline ABL1 â€associated congenital heart defects and skeletal malformations syndrome. Human Mutation, 2020, 41, 1738-1744.	2.5	10
64	A satellite-like sequence, representing a â€œclone gapâ€•in the human genome, was likely involved in the seeding of a novel centromere in macaque. Chromosoma, 2009, 118, 269-277.	2.2	9
65	Thermoelastic Deformations of Cylindrical Multi-Layered Shells Using a Direct Approach. Journal of Thermal Stresses, 2013, 36, 749-789.	2.0	8
66	Segregation of Incomplete Achromatopsia and Alopecia Due to PDE6H and LPAR6 Variants in a Consanguineous Family from Pakistan. Genes, 2016, 7, 41.	2.4	8
67	STIM1 R304W in mice causes subgingival hair growth and an increased fraction of trabecular bone. Cell Calcium, 2020, 85, 102110.	2.4	8
68	Inheritance of a terminal 7.1â€‰Mb 18p deletion flanked by a 2.3â€‰Mb duplication from a physically normal mother. American Journal of Medical Genetics, Part A, 2009, 149A, 2877-2881.	1.2	7
69	On the theory of loaded general cylindrical Cosserat elastic shells. International Journal of Solids and Structures, 2007, 44, 7399-7419.	2.7	6
70	On the Cosserat model for thin rods made of thermoelastic materials with voids. Discrete and Continuous Dynamical Systems - Series S, 2013, 6, 1473-1485.	1.1	6
71	DNA methylation episignature in Gabriele-de VriesÂsyndrome. Genetics in Medicine, 2022, 24, 905-914.	2.4	6
72	The Korn-type inequality in a Cosserat model for thin thermoelastic porous rods. Meccanica, 2012, 47, 789-794.	2.0	5

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73	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. BMC Medical Genetics, 2020, 21, 96.	2.1	5
74	ZBTB11 dysfunction: spectrum of brain abnormalities, biochemical signature and cellular consequences. Brain, 2022, 145, 2602-2616.	7.6	5
75	Minimum Energy Characterizations for the Solution of Saint-Venant's Problem in the Theory of Shells. Journal of Elasticity, 2005, 81, 179-204.	1.9	4
76	A de novo 6p interstitial deletion and a complex translocation involving chromosomes 2, 6, and 14 in a mildly developmentally delayed patient. American Journal of Medical Genetics, Part A, 2008, 146A, 3230-3233.	1.2	4
77	Thermal stresses in anisotropic cylindrical elastic shells. Mathematical Methods in the Applied Sciences, 2010, 33, 799-810.	2.3	3
78	A balanced de novo inv(7)(p14.3q22.3) disrupting PDE1C and ATXN7L1 in a 14-year old developmentally delayed boy. European Journal of Medical Genetics, 2013, 56, 361-364.	1.3	3
79	On the Bending Equations for Elastic Plates with Voids. Mathematics and Mechanics of Solids, 2007, 12, 40-57.	2.4	2
80	On the dynamic deformation of porous Cosserat linearâ€“thermoelastic shells. ZAMM Zeitschrift Fur Angewandte Mathematik Und Mechanik, 2008, 88, 74-78.	1.6	2
81	A girl with a neurodevelopmental syndrome, adducted thumbs and frequent infections caused by novel homozygous variant in DEAF1. Clinical Dysmorphology, 2020, 29, 107-110.	0.3	2
82	A partial trisomy 1q patient with a deletion 1q22 and an insertion 1(q42q44) into 1q22. American Journal of Medical Genetics, Part A, 2009, 149A, 290-293.	1.2	1
83	A de novo 15q13.2q13.3 deletion in a boy with an Angelman syndrome like phenotype. European Journal of Medical Genetics, 2010, 53, 221-224.	1.3	1
84	Closed-Form Saint-Venant Solutions in the Koiter Theory of Shells. Journal of Elasticity, 2020, 140, 149-169.	1.9	1
85	On a Problem of Truesdell for Anisotropic Elastic Shells. Analele Stiintifice Ale Universitatii Al I Cuza Din Iasi - Matematica, 2011, 57, .	0.2	0