Marco Castori

List of Publications by Year in descending order

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217 papers	8,064 citations	66234 42 h-index	64668 79 g-index
221 all docs	221 docs citations	221 times ranked	8492 citing authors

#	Article	IF	Citations
1	DNA methylation episignature testing improves molecular diagnosis of Mendelian chromatinopathies. Genetics in Medicine, 2022, 24, 51-60.	1.1	24
2	A novel complex genomic rearrangement affecting the KCNJ2 regulatory region causes a variant of Cooks syndrome. Human Genetics, 2022, 141, 217-227.	1.8	1
3	A novel homozygous variant in <scp>COX5A</scp> causes an attenuated phenotype with failure to thrive, lactic acidosis, hypoglycemia, and short stature. Clinical Genetics, 2022, 102, 56-60.	1.0	3
4	Transcriptome Analysis Reveals Altered Expression of Genes Involved in Hypoxia, Inflammation and Immune Regulation in Pdcd10-Depleted Mouse Endothelial Cells. Genes, 2022, 13, 961.	1.0	6
5	Generation of the induced pluripotent stem cell line UNIBSi017-A from an individual with cardiospondylocarpofacial syndrome and the MAP3K7 c.737-7A>G variant. Stem Cell Research, 2022, , 102837.	0.3	0
6	Expanding the phenotype associated to KMT2A variants: overlapping clinical signs between Wiedemannâ€“Steiner and Rubinsteinâ€“Taybi syndromes. European Journal of Human Genetics, 2021, 29, 88-98.	1.4	11
7	Whole Exome Sequencing Reveals a Novel AUTS2 In-Frame Deletion in a Boy with Global Developmental Delay, Absent Speech, Dysmorphic Features, and Cerebral Anomalies. Genes, 2021, 12, 229.	1.0	8
8	Improving clinical interpretation of five <scp>KRIT1</scp> and <scp>PDCD10</scp> intronic variants. Clinical Genetics, 2021, 99, 829-835.	1.0	1
9	Deconstructing and reconstructing joint hypermobility on an evo-devo perspective. Rheumatology, 2021, 60, 2537-2544.	0.9	5
10	Craniosynostosis is a feature of <scp>CHD7</scp>â€related <scp>CHARGE</scp> syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2160-2163.	0.7	2
11	Copy number variation analysis implicates novel pathways in patients with oculoâ€auriculoâ€vertebralâ€spectrum and congenital heart defects. Clinical Genetics, 2021, 100, 268-279.	1.0	9
12	Compound Heterozygosity for OTOA Truncating Variant and Genomic Rearrangement Cause Autosomal Recessive Sensorineural Hearing Loss in an Italian Family. Audiology Research, 2021, 11, 443-451.	0.8	0
13	Clinical presentation and molecular characterization of a novel patient with variant <scp>POC1A</scp>â€related syndrome. Clinical Genetics, 2021, 99, 540-546.	1.0	7
14	Review of clinical and molecular variability in autosomal recessive cutis laxa 2A. American Journal of Medical Genetics, Part A, 2021, 185, 955-965.	0.7	2
15	High rate of dyspareunia and probable vulvodynia in <scp>Ehlersâ€“Danlos</scp> syndromes and hypermobility spectrum disorders: An online survey. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 599-608.	0.7	11
16	Gonosomal Mosaicism for a Novel COL5A1 Pathogenic Variant in Classic Ehlers-Danlos Syndrome. Genes, 2021, 12, 1928.	1.0	3
17	Loss-of-function variants in exon 4 of TAB2 causeÂaÂrecognizable multisystem disorder with cardiovascular, facial, cutaneous, and musculoskeletalÂinvolvement. Genetics in Medicine, 2021, , .	1.1	1
18	Ehlers-Danlos Syndromes, Joint Hypermobility and Hypermobility Spectrum Disorders. Advances in Experimental Medicine and Biology, 2021, 1348, 207-233.	0.8	4

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19	A proposal of rehabilitative approach in the rare disease "De Barsy Syndrome": case report. Clinica Terapeutica, 2021, 171, e4-e7.	0.2	0
20	Application of the 2017 criteria for vascular Ehlersâ€Danlos syndrome in 50 patients ascertained according to the Villefranche nosology. Clinical Genetics, 2020, 97, 287-295.	1.0	7
21	Double missense mutations in cardiac myosinâ€binding protein C and myopalladin genes: A case report with diffuse coronary disease, complete atrioventricular block, and progression to dilated cardiomyopathy. Annals of Noninvasive Electrocardiology, 2020, 25, e12687.	0.5	7
22	COL1â€related overlap disorder: A novel connective tissue disorder incorporating the osteogenesis imperfecta/Ehlersâ€Danlos syndrome overlap. Clinical Genetics, 2020, 97, 396-406.	1.0	27
23	Novel TONSL variants cause SPONASTRIME dysplasia and associate with spontaneous chromosome breaks, defective cell proliferation and apoptosis. Human Molecular Genetics, 2020, 29, 3122-3131.	1.4	3
24	ACE2 gene variants may underlie interindividual variability and susceptibility to COVID-19 in the Italian population. European Journal of Human Genetics, 2020, 28, 1602-1614.	1.4	208
25	The Ehlersâ€“Danlos syndromes. Nature Reviews Disease Primers, 2020, 6, 64.	18.1	144
26	Pro-Fibrotic Phenotype in a Patient with Segmental Stiff Skin Syndrome via TGF-Î² Signaling Overactivation. International Journal of Molecular Sciences, 2020, 21, 5141.	1.8	9
27	Consensus based recommendations for diagnosis and medical management of Poland syndrome (sequence). Orphanet Journal of Rare Diseases, 2020, 15, 201.	1.2	17
28	Exon-Trapping Assay Improves Clinical Interpretation of COL11A1 and COL11A2 Intronic Variants in Stickler Syndrome Type 2 and Otospondylomegaepiphyseal Dysplasia. Genes, 2020, 11, 1513.	1.0	11
29	Novel Pathogenic Variants of the AIRE Gene in Two Autoimmune Polyendocrine Syndrome Type I Cases with Atypical Presentation: Role of the NGS in Diagnostic Pathway and Review of the Literature. Biomedicines, 2020, 8, 631.	1.4	2
30	Propranolol for familial cerebral cavernous malformation (Treat_CCM): study protocol for a randomized controlled pilot trial. Trials, 2020, 21, 401.	0.7	37
31	Rare Somatic MEN1 Gene Pathogenic Variant in a Patient Affected by Atypical Parathyroid Adenoma. International Journal of Endocrinology, 2020, 2020, 1-5.	0.6	4
32	The recurrent SETBP1 c.2608Gâ€‰>â€‰A, p.(Gly870Ser) variant in a patient with Schinzel-Giedion syndrome: an illustrative case of the utility of whole exome sequencing in a critically ill neonate. Italian Journal of Pediatrics, 2020, 46, 74.	1.0	6
33	Customised next-generation sequencing multigene panel to screen a large cohort of individuals with chromatin-related disorder. Journal of Medical Genetics, 2020, 57, 760-768.	1.5	15
34	Myotonic dystrophy type 1 cosegregating with autosomal dominant polycystic kidney disease type 2. Neurological Sciences, 2020, 41, 3761-3763.	0.9	0
35	A Private 16q24.2q24.3 Microduplication in a Boy with Intellectual Disability, Speech Delay and Mild Dysmorphic Features. Genes, 2020, 11, 707.	1.0	10
36	Insights into the molecular pathogenesis of cardiospondylocarpofacial syndrome: MAP3K7 c.737-7AÂ>ÂG variant alters the TGFÎ²-mediated Î±-SMA cytoskeleton assembly and autophagy. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2020, 1866, 165742.	1.8	7

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37	The splice c.1815G>A variant in KIAA0586 results in a phenotype bridging short-rib-polydactyly and oral-facial-digital syndrome. Medicine (United States), 2020, 99, e19169.	0.4	7
38	Compound Phenotype Due to Recessive Variants in LARP7 and OTOG Genes Disclosed by an Integrated Approach of SNP-Array and Whole Exome Sequencing. Genes, 2020, 11, 379.	1.0	3
39	Response to: Concern regarding classification of c.703G>A/p.Gly235Arg as a novel missense variant in KRIT1 gene. Human Mutation, 2020, 41, 1072-1074.	1.1	0
40	A new insight on postural tachycardia syndrome in 102 adults with hypermobile Ehlers-Danlos Syndrome/hypermobility spectrum disorder. Monaldi Archives for Chest Disease, 2020, 90, .	0.3	13
41	Joint hypermobility in children: a neglected sign needing more attention. Minerva Pediatrica, 2020, 72, 123-133.	2.6	6
42	TAB2 c.1398dup variant leads to haploinsufficiency and impairs extracellular matrix homeostasis. Human Mutation, 2019, 40, 1886-1898.	1.1	5
43	Molecular diagnostic workflow, clinical interpretation of sequence variants, and data repository procedures in 140 individuals with familial cerebral cavernous malformations. Human Mutation, 2019, 40, e24-e36.	1.1	3
44	Characterization of Two Novel Intronic Variants Affecting Splicing in FBN1-Related Disorders. Genes, 2019, 10, 442.	1.0	17
45	Mutational spectrum and clinical signatures in 114 families with hereditary multiple osteochondromas: insights into molecular properties of selected exostosin variants. Human Molecular Genetics, 2019, 28, 2133-2142.	1.4	12
46	A novel dominant-negative FGFR1 variant causes Hartsfield syndrome by deregulating RAS/ERK1/2 pathway. European Journal of Human Genetics, 2019, 27, 1113-1120.	1.4	12
47	Cardiac valvular Ehlersâ€Danlos syndrome is a wellâ€defined condition due to recessive null variants in COL1A2. American Journal of Medical Genetics, Part A, 2019, 179, 846-851.	0.7	15
48	Severity classes in adults with hypermobile Ehlersâ€“Danlos syndrome/hypermobility spectrum disorders: a pilot study of 105 Italian patients. Rheumatology, 2019, 58, 1722-1730.	0.9	22
49	Novel TNXB Variants in Two Italian Patients with Classical-Like Ehlers-Danlos Syndrome. Genes, 2019, 10, 967.	1.0	10
50	Primary muscle involvement in a 15â€yearâ€old girl with the recurrent homozygous c.362dupC variant in FKBP14. American Journal of Medical Genetics, Part A, 2019, 179, 317-321.	0.7	3
51	LTBP2â€related â€œMarfanâ€likeâ€•phenotype in two Roma/Gypsy subjects with the LTBP2 homozygous p.R299X variant. American Journal of Medical Genetics, Part A, 2019, 179, 104-112.	0.7	10
52	Italian validation of the functional difficulties questionnaire (FDQâ€9) and its correlation with major determinants of quality of life in adults with hypermobile Ehlersâ€“Danlos syndrome/hypermobility spectrum disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2019, 180, 25-34.	1.1	11
53	Facial comedonal acne in orofaciodigital syndrome type 1 caused by a novel frameshift variant in <scp>OFD</scp> 1 . Clinical and Experimental Dermatology, 2019, 44, 706-708.	0.6	2
54	A novel MAP3K7 splice mutation causes cardiospondylocarpofacial syndrome with features of hereditary connective tissue disorder. European Journal of Human Genetics, 2018, 26, 582-586.	1.4	14

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55	Hereditary palmoplantar keratodermas. Part I. Nonâ€syndromic palmoplantar keratodermas: classification, clinical and genetic features. Journal of the European Academy of Dermatology and Venereology, 2018, 32, 704-719.	1.3	47
56	Attention-deficit/hyperactivity disorder, joint hypermobility-related disorders and pain: expanding body-mind connections to the developmental age. ADHD Attention Deficit and Hyperactivity Disorders, 2018, 10, 163-175.	1.7	33
57	Hereditary palmoplantar keratodermas. Part <scp>II</scp>: syndromic palmoplantar keratodermas â€“ Diagnostic algorithm and principles of therapy. Journal of the European Academy of Dermatology and Venereology, 2018, 32, 899-925.	1.3	34
58	Variants in members of the cadherinâ€“catenin complex, CDH1 and CTNND1, cause blepharocheilodontic syndrome. European Journal of Human Genetics, 2018, 26, 210-219.	1.4	34
59	A recognizable systemic connective tissue disorder with polyvalvular heart dystrophy and dysmorphism associated with <scp>TAB2</scp> mutations. Clinical Genetics, 2018, 93, 126-133.	1.0	19
60	Ehlers-Danlos syndromes: state of the art on clinical practice guidelines. RMD Open, 2018, 4, e000790.	1.8	23
61	A novel mutation in CDH11, encoding cadherinâ€11, cause Branchioskeletogenital (Elsahyâ€Waters) syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2028-2033.	0.7	13
62	A single-center study on 140 patients with cerebral cavernous malformations: 28 new pathogenic variants and functional characterization of a PDCD10 large deletion. Human Mutation, 2018, 39, 1885-1900.	1.1	16
63	Exploring relationships between joint hypermobility and neurodevelopment in children (4â€“13 years) with hereditary connective tissue disorders and developmental coordination disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 546-556.	1.1	19
64	Pharmacological resources, diagnostic approach and coordination of care in joint hypermobility-related disorders. Expert Review of Clinical Pharmacology, 2018, 11, 689-703.	1.3	6
65	Biallelic variants in the ciliary gene TMEM67 cause RHYNS syndrome. European Journal of Human Genetics, 2018, 26, 1266-1271.	1.4	12
66	Clinical Relevance of Joint Hypermobility and Its Impact on Musculoskeletal Pain and Bone Mass. Current Osteoporosis Reports, 2018, 16, 333-343.	1.5	9
67	Genetic testing for vascular Ehlers-Danlos syndrome and other variants with fragility of the middle arteries. The EuroBiotech Journal, 2018, 2, 42-44.	0.5	0
68	Genetic testing for Marfan-like disorders. The EuroBiotech Journal, 2018, 2, 38-41.	0.5	0
69	Hypermobile Ehlersâ€“Danlos syndrome (a.k.a. Ehlersâ€“Danlos syndrome Type III and Ehlersâ€“Danlos) Tj ETQq1 1 0.784314 rgBT /Ov Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 48-69.	0.7	298
70	A framework for the classification of joint hypermobility and related conditions. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 148-157.	0.7	356
71	COL6A5variants in familial neuropathic chronic itch. Brain, 2017, 140, aww343.	3.7	25
72	Refining patterns of joint hypermobility, habitus, and orthopedic traits in joint hypermobility syndrome and Ehlersâ€“Danlos syndrome, hypermobility type. American Journal of Medical Genetics, Part A, 2017, 173, 914-929.	0.7	20

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73	Spectrum of mucocutaneous, ocular and facial features and delineation of novel presentations in 62 classical Ehlersâ€Danlos syndrome patients. Clinical Genetics, 2017, 92, 624-631.	1.0	26
74	The 2017 international classification of the Ehlersâ€“Danlos syndromes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 8-26.	0.7	1,163
75	De Novo Mutations in SLC25A24 Cause a Disorder Characterized by Early Aging, Bone Dysplasia, Characteristic Face, and Early Demise. American Journal of Human Genetics, 2017, 101, 844-855.	2.6	51
76	Contemporary approach to joint hypermobility and related disorders. Current Opinion in Pediatrics, 2017, 29, 640-649.	1.0	68
77	Posterior column ataxia with retinitis pigmentosa coexisting with sensoryâ€autonomic neuropathy and leukemia due to the homozygous p.Pro221Ser FLVCR1 mutation. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2017, 174, 732-739.	1.1	21
78	Ehlersâ€“Danlos syndrome with lethal cardiac valvular dystrophy in males carrying a novel splice mutation in FLNA. American Journal of Medical Genetics, Part A, 2017, 173, 169-176.	0.7	13
79	Identification of a second <scp>HOXA2</scp> nonsense mutation in a family with autosomal dominant nonâ€syndromic microtia and distinctive ear morphology. Clinical Genetics, 2017, 91, 774-779.	1.0	28
80	Orthostatic Intolerance and Postural Orthostatic Tachycardia Syndrome in Joint Hypermobility Syndrome/Ehlers-Danlos Syndrome, Hypermobility Type: Neurovegetative Dysregulation or Autonomic Failure?. BioMed Research International, 2017, 2017, 1-7.	0.9	28
81	Mutations in the Heme Exporter FLVCR1 Cause Sensory Neurodegeneration with Loss of Pain Perception. PLoS Genetics, 2016, 12, e1006461.	1.5	43
82	Clinical and molecular characterization of two patients with palmoplantar keratoderma-congenital alopecia syndrome type 2. Clinical and Experimental Dermatology, 2016, 41, 632-635.	0.6	8
83	Axial skeletogenesis in human autosomal aneuploidies: A radiographic study of 145 second trimester fetuses. American Journal of Medical Genetics, Part A, 2016, 170, 676-687.	0.7	11
84	Variability in a threeâ€generation family with pierre robin sequence, acampomelic campomelic dysplasia, and intellectual disability due to a novel â¼1 Mb deletion upstream of SOX9, and including KCNJ2 and KCNJ16. Birth Defects Research Part A: Clinical and Molecular Teratology, 2016, 106, 61-68.	1.6	13
85	Prediction and visualization data for the interpretation of sarcomeric and non-sarcomeric DNA variants found in patients with hypertrophic cardiomyopathy. Data in Brief, 2016, 7, 607-613.	0.5	0
86	Pain in Ehlers-Danlos syndromes: manifestations, therapeutic strategies and future perspectives. Expert Opinion on Orphan Drugs, 2016, 4, 1145-1158.	0.5	34
87	Clinical and molecular characterization of a boy with intellectual disability, facial dysmorphism, minor digital anomalies and a complex IL1RAPL1 intragenic rearrangement. European Journal of Paediatric Neurology, 2016, 20, 971-976.	0.7	8
88	Central sensitization as the mechanism underlying pain in joint hypermobility syndrome/Ehlersâ€“Danlos syndrome, hypermobility type. European Journal of Pain, 2016, 20, 1319-1325.	1.4	71
89	From the bedside to the bench and backwards: diagnostic approach and management of Ehlers-Danlos syndrome(s) in Italy. Journal of Medical Rehabilitation, 2016, 36, 9-27.	0.0	0
90	Small fiber neuropathy is a common feature of Ehlers-Danlos syndromes. Neurology, 2016, 87, 155-159.	1.5	90

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91	Molecular analysis of sarcomeric and non-sarcomeric genes in patients with hypertrophic cardiomyopathy. Gene, 2016, 577, 227-235.	1.0	26
92	TMJ replacement utilizing patient-fitted TMJ TJR devices in a re-ankylosis child. Journal of Cranio-Maxillo-Facial Surgery, 2016, 44, 493-499.	0.7	22
93	Early Mandibular Distraction to Relieve Robin Severe Airway Obstruction in Two Siblings with Lymphedemaâ€“Distichiasis Syndrome. Journal of Maxillofacial and Oral Surgery, 2016, 15, 384-389.	0.6	2
94	Transcriptome-Wide Expression Profiling in Skin Fibroblasts of Patients with Joint Hypermobility Syndrome/Ehlers-Danlos Syndrome Hypermobility Type. PLoS ONE, 2016, 11, e0161347.	1.1	40
95	Microcephaly, ectodermal dysplasia, multiple skeletal anomalies and distinctive facial appearance: Delineation of cerebroâ€dermatoâ€osseousâ€dysplasia. American Journal of Medical Genetics, Part A, 2015, 167, 842-851.	0.7	1
96	Ehlersâ€“Danlos syndrome(s) mimicking child abuse: Is there an impact on clinical practice?. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2015, 169, 289-292.	0.7	26
97	Phenotypic variability in developmental coordination disorder: Clustering of generalized joint hypermobility with attention deficit/hyperactivity disorder, atypical swallowing and narrative difficulties. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2015, 169, 117-122.	0.7	16
98	The Use of Piezosurgery in Cranial Surgery in Children. Journal of Craniofacial Surgery, 2015, 26, 840-842.	0.3	16
99	Nutritional Supplementation in Ehlers-Danlos Syndrome. , 2015, , 161-170.		1
100	Aortic dissection and stroke in a 37-year-old woman: discovering an emerging heritable connective tissue disorder. Internal and Emergency Medicine, 2015, 10, 165-170.	1.0	1
101	Gastrointestinal and nutritional issues in joint hypermobility syndrome/ehlersâ€“danlos syndrome, hypermobility type. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2015, 169, 54-75.	0.7	76
102	A study of migraine characteristics in joint hypermobility syndrome a.k.a. Ehlersâ€“Danlos syndrome, hypermobility type. Neurological Sciences, 2015, 36, 1417-1424.	0.9	37
103	Neurodevelopmental attributes of joint hypermobility syndrome/Ehlersâ€“Danlos syndrome, hypermobility type: Update and perspectives. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2015, 169, 107-116.	0.7	45
104	Generalized joint hypermobility, joint hypermobility syndrome and Ehlersâ€Danlos syndrome, hypermobility type. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2015, 169, 1-5.	0.7	33
105	Psychopathological manifestations of joint hypermobility and joint hypermobility syndrome/ Ehlersâ€“Danlos syndrome, hypermobility type: The link between connective tissue and psychological distress revised. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2015, 169, 97-106.	0.7	60
106	Spectrum of mucocutaneous manifestations in 277 patients with joint hypermobility syndrome/Ehlersâ€Danlos syndrome, hypermobility type. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2015, 169, 43-53.	0.7	30
107	Connective tissue, Ehlersâ€“Danlos syndrome(s), and head and cervical pain. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2015, 169, 84-96.	0.7	48
108	Oropharyngeal teratoma, oral duplication, cervical diplomyelia and anencephaly in a 22â€week fetus: A review of the craniofacial teratoma syndrome. Birth Defects Research Part A: Clinical and Molecular Teratology, 2015, 103, 554-566.	1.6	9

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109	An Additional Patient With 3q27.3 Microdeletion Syndrome. Journal of Child Neurology, 2015, 30, 500-504.	0.7	2
110	Foot Type Analysis Based on Electronic Pedobarography Data in Individuals with Joint Hypermobility Syndrome/Ehlers-Danlos Syndrome Hypermobility Type During Upright Standing. Journal of the American Podiatric Medical Association, 2014, 104, 588-593.	0.2	7
111	Unexpected association between joint hypermobility syndrome/Ehlersâ€“Danlos syndrome hypermobility type and obsessiveâ€“compulsive personality disorder. Rheumatology International, 2014, 34, 631-636.	1.5	30
112	Comparison of ultrasound and magnetic resonance imaging in the prenatal diagnosis of Apert syndrome: report of a case. Child's Nervous System, 2014, 30, 1445-8.	0.6	14
113	Towards a reâ€thinking of the clinical significance of generalized joint hypermobility, joint hypermobiity syndrome, and Ehlersâ€Danlos syndrome, hypermobility type. American Journal of Medical Genetics, Part A, 2014, 164, 588-590.	0.7	10
114	Heart rate, conduction and ultrasound abnormalities in adults with joint hypermobility syndrome/Ehlers-Danlos syndrome, hypermobility type. Clinical Rheumatology, 2014, 33, 981-987.	1.0	16
115	Late diagnosis of lateral meningocele syndrome in a 55â€yearâ€old woman with symptoms of joint instability and chronic musculoskeletal pain. American Journal of Medical Genetics, Part A, 2014, 164, 528-534.	0.7	16
116	Phenotype and genotype in Nicolaidesâ€“Baraitser syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 302-314.	0.7	66
117	Nosology and inheritance pattern(s) of joint hypermobility syndrome and Ehlersâ€Danlos syndrome, hypermobility type: A study of intrafamilial and interfamilial variability in 23 Italian pedigrees. American Journal of Medical Genetics, Part A, 2014, 164, 3010-3020.	0.7	70
118	Recommendations for anesthesia and perioperative management in patients with Ehlers-Danlos syndrome(s). Orphanet Journal of Rare Diseases, 2014, 9, 109.	1.2	83
119	Novel SMAD4 mutation causing Myhre syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 1835-1840.	0.7	29
120	A 22-Week-Old Fetus with Nager Syndrome and Congenital Diaphragmatic Hernia due to a Novel <i>SF3B4</i> Mutation. Molecular Syndromology, 2014, 5, 241-244.	0.3	15
121	Fast and Early Mandibular Osteogenetic Distraction in a 24-Day-Old Female Newborn With Larsen Syndrome. Journal of Craniofacial Surgery, 2014, 25, e304-e307.	0.3	1
122	Joint hypermobility syndrome/Ehlersâ€“Danlos syndrome hypermobility type: constructing a rehabilitative approach. International Journal of Clinical Rheumatology, 2014, 9, 103-106.	0.3	3
123	Neurological manifestations of Ehlers-Danlos syndrome(s): A review. Iranian Journal of Neurology, 2014, 13, 190-208.	0.5	45
124	Diabetic Embryopathy: A Developmental Perspective from Fertilization to Adulthood. Molecular Syndromology, 2013, 4, 74-86.	0.3	37
125	Ehlersâ€“Danlos syndrome hypermobility type: a possible unifying concept for various functional somatic syndromes. Rheumatology International, 2013, 33, 819-821.	1.5	17
126	Genotypeâ€“phenotype spectrum of PYCR1-related autosomal recessive cutis laxa. Molecular Genetics and Metabolism, 2013, 110, 352-361.	0.5	57

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127	Prenatal diagnosis and post-mortem examination in a fetus with thrombocytopenia-absent radius (TAR) syndrome due to compound heterozygosity for a 1q21.1 microdeletion and a RBM8A hypomorphic allele: a case report. BMC Research Notes, 2013, 6, 376.	0.6	24
128	Clinical and genetic study of two patients with Zimmermannâ€“Laband syndrome and literature review. European Journal of Medical Genetics, 2013, 56, 570-576.	0.7	32
129	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.	0.7	1
130	Use of the Gait Profile Score for the evaluation of patients with joint hypermobility syndrome/Ehlersâ€“Danlos syndrome hypermobility type. Research in Developmental Disabilities, 2013, 34, 4280-4285.	1.2	43
131	Entrapment neuropathies and polyneuropathies in joint hypermobility syndrome/Ehlersâ€“Danlos syndrome. Clinical Neurophysiology, 2013, 124, 1689-1694.	0.7	32
132	Novel mutations of the <scp>PRKAR1A</scp> gene inÂpatients with acrodysostosis. Clinical Genetics, 2013, 84, 531-538.	1.0	27
133	Evaluation of Kinesiophobia and Its Correlations with Pain and Fatigue in Joint Hypermobility Syndrome/Ehlers-Danlos Syndrome Hypermobility Type. BioMed Research International, 2013, 2013, 1-7.	0.9	60
134	Reâ€•writing the natural history of pain and related symptoms in the joint hypermobility syndrome/Ehlersâ€“Danlos syndrome, hypermobility type. American Journal of Medical Genetics, Part A, 2013, 161, 2989-3004.	0.7	126
135	Joint hypermobility syndrome (a.k.a. Ehlers-Danlos Syndrome, Hypermobility Type): an updated critique. Giornale Italiano Di Dermatologia E Venereologia, 2013, 148, 13-36.	0.8	14
136	Ehlers-Danlos Syndrome, Hypermobility Type: An Underdiagnosed Hereditary Connective Tissue Disorder with Mucocutaneous, Articular, and Systemic Manifestations. ISRN Dermatology, 2012, 2012, 1-22.	1.9	159
137	PRKAR1AandPDE4DMutations Cause Acrodysostosis but Two Distinct Syndromes with or without GPCR-Signaling Hormone Resistance. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E2328-E2338.	1.8	100
138	Surgical Recommendations in Ehlers-Danlos Syndrome(s) Need Patient Classification: The Example of Ehlers-Danlos Syndrome Hypermobility Type (a.k.a. Joint Hypermobility Syndrome). Digestive Surgery, 2012, 29, 453-455.	0.6	14
139	Genetic skin diseases predisposing to basal cell carcinoma. European Journal of Dermatology, 2012, 22, 299-309.	0.3	71
140	Relationship between fatigue and gait abnormality in Joint Hypermobility Syndrome/Ehlers-Danlos Syndrome Hypermobility type. Research in Developmental Disabilities, 2012, 33, 1914-1918.	1.2	30
141	Heterozygous missense mutations in SMARCA2 cause Nicolaides-Baraitser syndrome. Nature Genetics, 2012, 44, 445-449.	9.4	207
142	Gynecologic and obstetric implications of the joint hypermobility syndrome (a.k.a. Ehlersâ€“Danlos) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5 158A, 2176-2182.	0.7	78
143	Ocular Features in Joint Hypermobility Syndrome/Ehlers-Danlos Syndrome Hypermobility Type: A Clinical and In Vivo Confocal Microscopy Study. American Journal of Ophthalmology, 2012, 154, 593-600.e1.	1.7	47
144	De Barsy Syndrome: A genetically heterogeneous autosomal recessive cutis laxa syndrome related to P5CS and PYCR1 dysfunction. American Journal of Medical Genetics, Part A, 2012, 158A, 927-931.	0.7	37

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145	Adult presentation of arterial tortuosity syndrome in a 51â€yearâ€old woman with a novel homozygous c.1411+1G>A mutation in the SLC2A10 gene. American Journal of Medical Genetics, Part A, 2012, 158A, 1164-1169.	0.7	25
146	Management of pain and fatigue in the joint hypermobility syndrome (a.k.a. Ehlersâ€“Danlos syndrome,) Tj ETQq0 0 0 rgBT /Overlock 10 Medical Genetics, Part A, 2012, 158A, 2055-2070.	0.7	124
147	Analysis of the miR-34a locus in 62 patients with familial cutaneous melanoma negative for CDKN2A/CDK4 screening. Familial Cancer, 2012, 11, 201-208.	0.9	10
148	Evaluation of lower limb disability in joint hypermobility syndrome. Rheumatology International, 2012, 32, 2577-2581.	1.5	10
149	Whorled hairless nevus of the scalp, linear hyperpigmentation, and telangiectatic nevi of the lower limbs: A novel variant of the â€œphacomatosis complexâ€. American Journal of Medical Genetics, Part A, 2012, 158A, 445-449.	0.7	1
150	Growth in Distal Arthrogryposes. , 2012, , 2265-2280.		0
151	Clinical features predicting identification of CDKN2A mutations in Italian patients with familial cutaneous melanoma. Cancer Epidemiology, 2011, 35, e116-e120.	0.8	24
152	Gait strategy in patients with Ehlersâ€“Danlos syndrome hypermobility type: A kinematic and kinetic evaluation using 3D gait analysis. Research in Developmental Disabilities, 2011, 32, 1663-1668.	1.2	46
153	Two families confirm SchÃ¶pf-Schulz-Passarge syndrome as a discrete entity within the WNT10A phenotypic spectrum. Clinical Genetics, 2011, 79, 92-95.	1.0	20
154	Pachydermodactyly with mild features of heritable connective tissue disorder and no sign of emotional distress. Clinical and Experimental Dermatology, 2011, 36, 690-692.	0.6	3
155	Novel mutations affecting LRP5 splicing in patients with osteoporosis-pseudoglioma syndrome (OPPG). European Journal of Human Genetics, 2011, 19, 875-881.	1.4	48
156	Neuropathic Pain Is a Common Feature in Ehlers-Danlos Syndrome. Journal of Pain and Symptom Management, 2011, 41, e2-e4.	0.6	51
157	The nosology of Richieriâ€Costa/Guionâ€Almeida syndrome(s). American Journal of Medical Genetics, Part A, 2011, 155, 398-402.	0.7	1
158	Early ultrasound suspect of thanatophoric dysplasia followed by first trimester molecular diagnosis. American Journal of Medical Genetics, Part A, 2011, 155, 1756-1758.	0.7	4
159	Screening for celiac disease in the joint hypermobility syndrome/Ehlersâ€“Danlos syndrome hypermobility type. American Journal of Medical Genetics, Part A, 2011, 155, 2314-2316.	0.7	28
160	Monozygotic twin discordance for phacomatosis cesioflammea further supports the postâ€zygotic mutation hypothesis. American Journal of Medical Genetics, Part A, 2011, 155, 2253-2256.	0.7	7
161	Reassessment of oral frenula in Ehlersâ€“Danlos syndrome: A study of 32 patients with the hypermobility type. American Journal of Medical Genetics, Part A, 2011, 155, 3157-3159.	0.7	12
162	Functional characterization of a novel TP63 mutation in a family with overlapping features of Rappâ€Hodgkin/AEC/ADULT syndromes. American Journal of Medical Genetics, Part A, 2011, 155, 3104-3109.	0.7	21

#	Article	IF	Citations
163	Evaluation of balance and improvement of proprioception by repetitive muscle vibration in a 15â€yearâ€old girl with joint hypermobility syndrome. Arthritis Care and Research, 2011, 63, 775-779.	1.5	27
164	AXIN2 germline mutations are rare in familial melanoma. Genes Chromosomes and Cancer, 2011, 50, 370-373.	1.5	8
165	Molecular characterization of 11 Italian patients with Darier Disease. European Journal of Dermatology, 2011, 21, 334-338.	0.3	12
166	Chronic fatigue syndrome is commonly diagnosed in patients with Ehlers-Danlos syndrome hypermobility type/joint hypermobility syndrome. Clinical and Experimental Rheumatology, 2011, 29, 597-8.	0.4	26
167	Symptom and joint mobility progression in the joint hypermobility syndrome (Ehlers-Danlos syndrome,) Tj ETQq1 1 0.784314 rgBT /Ov	0.4	57
168	Holoprosencephalyâ€diencephalic hamartoma: Sequence or pleiotropy?. American Journal of Medical Genetics, Part A, 2010, 152A, 264-266.	0.7	2
169	Systematized organoid epidermal nevus with eccrine differentiation, multiple facial and oral congenital scars, gingival synechiae, and blepharophimosis: A novel epidermal nevus syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 25-31.	0.7	13
170	Natural history and manifestations of the hypermobility type Ehlersâ€“Danlos syndrome: A pilot study on 21 patients. American Journal of Medical Genetics, Part A, 2010, 152A, 556-564.	0.7	172
171	Sirenomelia and VACTERL association in the offspring of a woman with diabetes. American Journal of Medical Genetics, Part A, 2010, 152A, 1803-1807.	0.7	28
172	Palmoplantar keratoderma, pseudoâ€ainhum, and universal atrichia: A new patient and review of the palmoplantar keratodermaâ€congenital alopecia syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 2043-2047.	0.7	17
173	Ehlersâ€“Danlos syndrome hypermobility type and the excess of affected females: Possible mechanisms and perspectives. American Journal of Medical Genetics, Part A, 2010, 152A, 2406-2408.	0.7	79
174	Elsahyâ€“Waters syndrome: Evidence for autosomal recessive inheritance. American Journal of Medical Genetics, Part A, 2010, 152A, 2810-2815.	0.7	12
175	Quality of life in the classic and hypermobility types of ElhersâEDanlos syndrome. Annals of Neurology, 2010, 67, 145-146.	2.8	38
176	Lethal autosomal recessive epidermolytic ichthyosis due to a novel donor splice-site mutation inKRT10. British Journal of Dermatology, 2010, 162, 1384-1387.	1.4	17
177	Novel and recurrent p14^{ARF} mutations in Italian familial melanoma. Clinical Genetics, 2010, 77, 581-586.	1.0	18
178	A Patient with Unilateral Tibial Aplasia and Accessory Scrotum: A Pure Coincidence or Nonfortuitous Association?. Case Reports in Medicine, 2010, 2010, 1-4.	0.3	4
179	Jejunal atresia and anterior chamber anomalies: Further delineation of the StrÃ¸mme syndrome. European Journal of Medical Genetics, 2010, 53, 149-152.	0.7	5
180	Palmoplantar keratoderma in keratosis follicularis spinulosa decalvans. European Journal of Dermatology, 2010, 20, 850-2.	0.3	2

#	Article	IF	Citations
181	Natural history of TFR2â€related hereditary hemochromatosis in a 47â€yrâ€old Italian patient. European Journal of Haematology, 2009, 83, 494-496.	1.1	5
182	Paradoxical association of extensive nevus flammeus together with unilateral lower limb and breast hypoplasia. American Journal of Medical Genetics, Part A, 2009, 149A, 266-267.	0.7	2
183	Juvenile macular dystrophy and forearm pronationâ€supination restriction presenting with features of distal arthrogryposis type 5. American Journal of Medical Genetics, Part A, 2009, 149A, 482-486.	0.7	7
184	Fontaineâ€“Farriaux syndrome: A recognizable craniosynostosis syndrome with nail, skeletal, abdominal, and central nervous system anomalies. American Journal of Medical Genetics, Part A, 2009, 149A, 2193-2199.	0.7	15
185	Darier disease, multiple bone cysts, and aniridia due to double de novo heterozygous mutations in ATP2A2 and PAX6. American Journal of Medical Genetics, Part A, 2009, 149A, 1768-1772.	0.7	7
186	NovelCTSCmutations in a patient with Papillon-LefÃ¨vre syndrome with recurrent pyoderma and minimal oral and palmoplantar involvement. British Journal of Dermatology, 2009, 160, 881-883.	1.4	13
187	Novel and recurrent germline LEMD3 mutations causing Buschkeâ€“Ollendorff syndrome and osteopoikilosis but not isolated melorheostosis. Clinical Genetics, 2009, 75, 556-561.	1.0	54
188	Trisomic rescue causing reduction to homozygosity for a novel ABCA12 mutation in harlequin ichthyosis. Clinical Genetics, 2009, 76, 392-397.	1.0	21
189	Clinical and genetic heterogeneity in keratosis follicularis spinulosa decalvans. European Journal of Medical Genetics, 2009, 52, 53-58.	0.7	28
190	Bazexâ€“DuprÃ©â€“Christol syndrome: An ectodermal dysplasia with skin appendage neoplasms. European Journal of Medical Genetics, 2009, 52, 250-255.	0.7	22
191	A novel heterozygous SOX2 mutation causing anophthalmia/microphthalmia with genital anomalies. European Journal of Medical Genetics, 2009, 52, 273-276.	0.7	4
192	VACTERL association and maternal diabetes: A possible causal relationship?. Birth Defects Research Part A: Clinical and Molecular Teratology, 2008, 82, 169-172.	1.6	35
193	Phacomatosis cesioflammea with unilateral lipohypoplasia. American Journal of Medical Genetics, Part A, 2008, 146A, 492-495.	0.7	13
194	Tibial developmental field defect is the most common lower limb malformation pattern in VACTERL association. American Journal of Medical Genetics, Part A, 2008, 146A, 1259-1266.	0.7	24
195	Complete maternal isodisomy causing reduction to homozygosity for a novel LAMB3 mutation in Herlitz junctional epidermolysis bullosa. Journal of Dermatological Science, 2008, 51, 58-61.	1.0	15
196	A rare cause of syndromic hypotrichosis: Nicolaides-Baraitser syndrome. Journal of the American Academy of Dermatology, 2008, 59, S92-S98.	0.6	4
197	Association of segmental neurofibromatosis 1 and oculo-auriculo-vertebral spectrum in a 24-year-old female. European Journal of Dermatology, 2008, 18, 22-5.	0.3	7
198	SchÃ¶pf-Schulz-Passarge Syndrome: Further Delineation of the Phenotype and Genetic Considerations. Acta Dermato-Venereologica, 2008, 88, 607-612.	0.6	39

#	Article	IF	Citations
199	Pai syndrome: First patient with agenesis of the corpus callosum and literature review. Birth Defects Research Part A: Clinical and Molecular Teratology, 2007, 79, 673-679.	1.6	18
200	A novel patient with Cooks syndrome supports splitting from â€œclassicâ€•brachydactyly type B. American Journal of Medical Genetics, Part A, 2007, 143A, 195-199.	0.7	5
201	Reassessment of holoprosencephalyâ€“diencephalic hamartoblastoma (HDH) association. American Journal of Medical Genetics, Part A, 2007, 143A, 277-284.	0.7	8
202	A triploid fetus further expands etiological heterogeneity in holoprosencephaly-diencephalic hamartoblastoma. American Journal of Medical Genetics, Part A, 2007, 143A, 1391-1393.	0.7	1
203	Syndromic craniosynostosis due to complex chromosome 5 rearrangement and MSX2 gene triplication. American Journal of Medical Genetics, Part A, 2007, 143A, 2937-2943.	0.7	31
204	Delayed diagnosis of dyskeratosis congenita in a 40-year-old woman with multiple head and neck squamous cell carcinomas. British Journal of Dermatology, 2007, 156, 406-408.	1.4	9
205	Herlitz junctional epidermolysis bullosa: laminin-5 mutational profile and carrier frequency in the Italian population. British Journal of Dermatology, 2007, 158, 071004160508001-???.	1.4	24
206	Reticulate Vascular Lesions and a Large Head. Pediatric Dermatology, 2007, 24, 555-556.	0.5	4
207	Mutations in CEP290, which encodes a centrosomal protein, cause pleiotropic forms of Joubert syndrome. Nature Genetics, 2006, 38, 623-625.	9.4	368
208	Hypochondrogenesis. Pediatric Radiology, 2006, 36, 460-461.	1.1	4
209	Antenatal presentation of the oculo-auriculo-vertebral spectrum (OAVS). American Journal of Medical Genetics, Part A, 2006, 140A, 1573-1579.	0.7	34
210	AHI1gene mutations cause specific forms of Joubert syndrome-related disorders. Annals of Neurology, 2006, 59, 527-534.	2.8	125
211	Pachydermoperiostosis: an update. Clinical Genetics, 2005, 68, 477-486.	1.0	190
212	Majewski osteodysplastic primordial dwarfism type II (MOPD II) complicated by stroke: Clinical report and review of cerebral vascular anomalies. American Journal of Medical Genetics, Part A, 2005, 139A, 212-215.	0.7	52
213	Distinguishing the four genetic causes of jouberts syndrome-related disorders. Annals of Neurology, 2005, 57, 513-519.	2.8	104
214	NPHP1 gene deletion is a rare cause of Joubert syndrome related disorders. Journal of Medical Genetics, 2005, 42, e9-e9.	1.5	93
215	A Novel Locus for Autosomal Dominant Cone and Coneâ€“Rod Dystrophies Maps to the 6p Gene Cluster of Retinal Dystrophies. , 2005, 46, 3539.		2
216	A locus for autosomal dominant keratoconus maps to human chromosome 3p14-q13. Journal of Medical Genetics, 2004, 41, 188-192.	1.5	118

#	Article	IF	Citations
217	Role of the dopamine D5 receptor (DRD5) as a susceptibility gene for cervical dystonia. Journal of Neurology, Neurosurgery and Psychiatry, 2003, 74, 665-666.	0.9	30