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	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
2	Mutations in CEP290, which encodes a centrosomal protein, cause pleiotropic forms of Joubert syndrome. Nature Genetics, 2006, 38, 623-625.	9.4	368
3	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
4	Hypermobile Ehlersâ€“Danlos syndrome (a.k.a. Ehlersâ€“Danlos syndrome Type III and Ehlersâ€“Danlos) Tj ETQq0 0 0 rgBT /Overlock 10 Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 48-69.	0.7	298
5	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
6	Heterozygous missense mutations in SMARCA2 cause Nicolaides-Baraitser syndrome. Nature Genetics, 2012, 44, 445-449.	9.4	207
7	Pachydermoperiostosis: an update. Clinical Genetics, 2005, 68, 477-486.	1.0	190
8	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
9	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
10	The Ehlersâ€“Danlos syndromes. Nature Reviews Disease Primers, 2020, 6, 64.	18.1	144
11	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
12	AHI1gene mutations cause specific forms of Joubert syndrome-related disorders. Annals of Neurology, 2006, 59, 527-534.	2.8	125
13	Management of pain and fatigue in the joint hypermobility syndrome (a.k.a. Ehlersâ€“Danlos syndrome,) Tj ETQq1 1 0.784314 rgBT /O Medical Genetics, Part A, 2012, 158A, 2055-2070.	0.7	124
14	A locus for autosomal dominant keratoconus maps to human chromosome 3p14-q13. Journal of Medical Genetics, 2004, 41, 188-192.	1.5	118
15	Distinguishing the four genetic causes of jouberts syndrome-related disorders. Annals of Neurology, 2005, 57, 513-519.	2.8	104
16	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
17	NPHP1 gene deletion is a rare cause of Joubert syndrome related disorders. Journal of Medical Genetics, 2005, 42, e9-e9.	1.5	93
18	Small fiber neuropathy is a common feature of Ehlers-Danlos syndromes. Neurology, 2016, 87, 155-159.	1.5	90

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19	Recommendations for anesthesia and perioperative management in patients with Ehlers-Danlos syndrome(s). Orphanet Journal of Rare Diseases, 2014, 9, 109.	1.2	83
20	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
21	Gynecologic and obstetric implications of the joint hypermobility syndrome (a.k.a. Ehlersâ€“Danlos) Tj ETQq1 1 0.784314 rgBT /Overlo 158A, 2176-2182.	0.7	78
22	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
23	Genetic skin diseases predisposing to basal cell carcinoma. European Journal of Dermatology, 2012, 22, 299-309.	0.3	71
24	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
25	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
26	Contemporary approach to joint hypermobility and related disorders. Current Opinion in Pediatrics, 2017, 29, 640-649.	1.0	68
27	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
28	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
29	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
30	Genotypeâ€“phenotype spectrum of PYCR1-related autosomal recessive cutis laxa. Molecular Genetics and Metabolism, 2013, 110, 352-361.	0.5	57
31	Symptom and joint mobility progression in the joint hypermobility syndrome (Ehlers-Danlos syndrome,) Tj ETQq1 1 0.784314 rgBT /Ov	0.4	57
32	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
33	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
34	Neuropathic Pain Is a Common Feature in Ehlers-Danlos Syndrome. Journal of Pain and Symptom Management, 2011, 41, e2-e4.	0.6	51
35	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
36	Novel mutations affecting LRP5 splicing in patients with osteoporosis-pseudoglioma syndrome (OPPG). European Journal of Human Genetics, 2011, 19, 875-881.	1.4	48

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37	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
38	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
39	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
40	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
41	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
42	Neurological manifestations of Ehlers-Danlos syndrome(s): A review. Iranian Journal of Neurology, 2014, 13, 190-208.	0.5	45
43	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
44	Mutations in the Heme Exporter FLVCR1 Cause Sensory Neurodegeneration with Loss of Pain Perception. PLoS Genetics, 2016, 12, e1006461.	1.5	43
45	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
46	SchÃ¶pf-Schulz-Passarge Syndrome: Further Delineation of the Phenotype and Genetic Considerations. Acta Dermato-Venereologica, 2008, 88, 607-612.	0.6	39
47	Quality of life in the classic and hypermobility types of ElhersâEDanlos syndrome. Annals of Neurology, 2010, 67, 145-146.	2.8	38
48	Diabetic Embryopathy: A Developmental Perspective from Fertilization to Adulthood. Molecular Syndromology, 2013, 4, 74-86.	0.3	37
49	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
50	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
51	Propranolol for familial cerebral cavernous malformation (Treat_CCM): study protocol for a randomized controlled pilot trial. Trials, 2020, 21, 401.	0.7	37
52	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
53	Antenatal presentation of the oculo-auriculo-vertebral spectrum (OAVS). American Journal of Medical Genetics, Part A, 2006, 140A, 1573-1579.	0.7	34
54	Pain in Ehlers-Danlos syndromes: manifestations, therapeutic strategies and future perspectives. Expert Opinion on Orphan Drugs, 2016, 4, 1145-1158.	0.5	34

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55	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
56	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
57	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
58	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
59	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
60	Entrapment neuropathies and polyneuropathies in joint hypermobility syndrome/Ehlersâ€“Danlos syndrome. Clinical Neurophysiology, 2013, 124, 1689-1694.	0.7	32
61	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
62	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
63	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
64	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
65	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
66	Novel SMAD4 mutation causing Myhre syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 1835-1840.	0.7	29
67	Clinical and genetic heterogeneity in keratosis follicularis spinulosa decalvans. European Journal of Medical Genetics, 2009, 52, 53-58.	0.7	28
68	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
69	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
70	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
71	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
72	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

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73	Novel mutations of the <scp>PRKAR1A</scp> gene inÂpatients with acrodysostosis. Clinical Genetics, 2013, 84, 531-538.	1.0	27
74	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
75	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
76	Molecular analysis of sarcomeric and non-sarcomeric genes in patients with hypertrophic cardiomyopathy. Gene, 2016, 577, 227-235.	1.0	26
77	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
78	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
79	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
80	COL6A5variants in familial neuropathic chronic itch. Brain, 2017, 140, aww343.	3.7	25
81	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
82	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
83	Clinical features predicting identification of CDKN2A mutations in Italian patients with familial cutaneous melanoma. Cancer Epidemiology, 2011, 35, e116-e120.	0.8	24
84	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
85	DNA methylation episignature testing improves molecular diagnosis of Mendelian chromatinopathies. Genetics in Medicine, 2022, 24, 51-60.	1.1	24
86	Ehlers-Danlos syndromes: state of the art on clinical practice guidelines. RMD Open, 2018, 4, e000790.	1.8	23
87	Bazexâ€“DuprÃ©â€“Christol syndrome: An ectodermal dysplasia with skin appendage neoplasms. European Journal of Medical Genetics, 2009, 52, 250-255.	0.7	22
88	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
89	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
90	Trisomic rescue causing reduction to homozygosity for a novel ABCA12 mutation in harlequin ichthyosis. Clinical Genetics, 2009, 76, 392-397.	1.0	21

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91	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
92	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
93	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
94	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
95	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
96	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
97	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
98	Novel and recurrent p14^{ARF} mutations in Italian familial melanoma. Clinical Genetics, 2010, 77, 581-586.	1.0	18
99	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
100	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
101	Ehlersâ€“Danlos syndrome hypermobility type: a possible unifying concept for various functional somatic syndromes. Rheumatology International, 2013, 33, 819-821.	1.5	17
102	Characterization of Two Novel Intronic Variants Affecting Splicing in FBN1-Related Disorders. Genes, 2019, 10, 442.	1.0	17
103	Consensus based recommendations for diagnosis and medical management of Poland syndrome (sequence). Orphanet Journal of Rare Diseases, 2020, 15, 201.	1.2	17
104	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
105	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
106	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
107	The Use of Piezosurgery in Cranial Surgery in Children. Journal of Craniofacial Surgery, 2015, 26, 840-842.	0.3	16
108	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

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109	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
110	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
111	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
112	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
113	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
114	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
115	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
116	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
117	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
118	Phacomatosis cesioflammea with unilateral lipohypoplasia. American Journal of Medical Genetics, Part A, 2008, 146A, 492-495.	0.7	13
119	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
120	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
121	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
122	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
123	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
124	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
125	Elsahyâ€“Waters syndrome: Evidence for autosomal recessive inheritance. American Journal of Medical Genetics, Part A, 2010, 152A, 2810-2815.	0.7	12
126	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

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127	Biallelic variants in the ciliary gene TMEM67 cause RHYNS syndrome. European Journal of Human Genetics, 2018, 26, 1266-1271.	1.4	12
128	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
129	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
130	Molecular characterization of 11 Italian patients with Darier Disease. European Journal of Dermatology, 2011, 21, 334-338.	0.3	12
131	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
132	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
133	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
134	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
135	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
136	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
137	Evaluation of lower limb disability in joint hypermobility syndrome. Rheumatology International, 2012, 32, 2577-2581.	1.5	10
138	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
139	Novel TNXB Variants in Two Italian Patients with Classical-Like Ehlers-Danlos Syndrome. Genes, 2019, 10, 967.	1.0	10
140	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
141	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
142	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
143	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
144	Clinical Relevance of Joint Hypermobility and Its Impact on Musculoskeletal Pain and Bone Mass. Current Osteoporosis Reports, 2018, 16, 333-343.	1.5	9

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145	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
146	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
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