Laura Mazzanti

List of Publications by Year
in descending order

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62 papers	4,063 citations	159358 30 h-index	118652 62 g-index
65 all docs	65 docs citations	65 times ranked	5544 citing authors

#	Article	IF	Citations
1	Gain-of-function RAF1 mutations cause Noonan and LEOPARD syndromes with hypertrophic cardiomyopathy. Nature Genetics, 2007, 39, 1007-1012.	9.4	624
2	Mutation of SHOC2 promotes aberrant protein N-myristoylation and causes Noonan-like syndrome with loose anagen hair. Nature Genetics, 2009, 41, 1022-1026.	9.4	358
3	A restricted spectrum of NRAS mutations causes Noonan syndrome. Nature Genetics, 2010, 42, 27-29.	9.4	271
4	GermlineBRAFmutations in Noonan, LEOPARD, and cardiofaciocutaneous syndromes: Molecular diversity and associated phenotypic spectrum. Human Mutation, 2009, 30, 695-702.	1.1	251
5	Molecular and Clinical Analyses of Greig Cephalopolysyndactyly and Pallister-Hall Syndromes: Robust Phenotype Prediction from the Type and Position of GLI3 Mutations. American Journal of Human Genetics, 2005, 76, 609-622.	2.6	248
6	Congenital heart disease in patients with Turnerâ€™s syndrome. Journal of Pediatrics, 1998, 133, 688-692.	0.9	233
7	Heterozygous Germline Mutations in the CBL Tumor-Suppressor Gene Cause a Noonan Syndrome-like Phenotype. American Journal of Human Genetics, 2010, 87, 250-257.	2.6	221
8	Prevalence and Clinical Picture of Celiac Disease in Turner Syndrome. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 5495-5498.	1.8	161
9	On the nosology and pathogenesis of Wolfâ€“Hirschhorn syndrome: Genotypeâ€“phenotype correlation analysis of 80 patients and literature review. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2008, 148C, 257-269.	0.7	143
10	SOS1 mutations in Noonan syndrome: molecular spectrum, structural insights on pathogenic effects, and genotype-phenotype correlations. Human Mutation, 2011, 32, 760-772.	1.1	97
11	Gonadoblastoma in Turner syndrome and Y-chromosome-derived material. American Journal of Medical Genetics, Part A, 2005, 135A, 150-154.	0.7	87
12	Molecular Analysis, Pathogenic Mechanisms, and Readthrough Therapy on a Large Cohort of<scp>K</scp>abuki Syndrome Patients. Human Mutation, 2014, 35, 841-850.	1.1	87
13	Characteristics of a nationwide cohort of patients presenting with isolated hypogonadotropic hypogonadism (IHH). European Journal of Endocrinology, 2018, 178, 23-32.	1.9	84
14	Loss-of-function variants of SETD5 cause intellectual disability and the core phenotype of microdeletion 3p25.3 syndrome. European Journal of Human Genetics, 2015, 23, 753-760.	1.4	73
15	Noonan-like syndrome with loose anagen hair: A new syndrome?. American Journal of Medical Genetics Part A, 2003, 118A, 279-286.	2.4	72
16	Recurrent Mutations in the Basic Domain of TWIST2 Cause Ablepharon Macrostomia and Barber-Say Syndromes. American Journal of Human Genetics, 2015, 97, 99-110.	2.6	61
17	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	2.6	48
18	Screening of PRKAR1A and PDE4D in a Large Italian Series of Patients Clinically Diagnosed With Albright Hereditary Osteodystrophy and/or Pseudohypoparathyroidism. Journal of Bone and Mineral Research, 2016, 31, 1215-1224.	3.1	47

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19	Pelvic ultrasonography in patients with Turner syndrome: Age-related findings in different karyotypes. Journal of Pediatrics, 1997, 131, 135-140.	0.9	45
20	Hearing loss in Turner syndrome: Results of a multicentric study. Journal of Endocrinological Investigation, 2008, 31, 779-783.	1.8	44
21	Quality of Life and Psychological Adjustment of Women Living with 46,XY Differences of Sex Development. Journal of Sexual Medicine, 2015, 12, 1440-1449.	0.3	44
22	Effects of sport (football) on growth auxological, anthropometric and hormonal aspects. European Journal of Applied Physiology and Occupational Physiology, 1990, 61, 149-158.	1.2	42
23	Epilepsy in Mowatâ€“Wilson syndrome: Delineation of the electroclinical phenotype. American Journal of Medical Genetics, Part A, 2013, 161, 273-284.	0.7	42
24	The Discovery of a LEMD2-Associated Nuclear Envelopathy with Early Progeroid Appearance Suggests Advanced Applications for AI-Driven Facial Phenotyping. American Journal of Human Genetics, 2019, 104, 749-757.	2.6	41
25	Loss of function of the E3 ubiquitin-protein ligase UBE3B causes Kaufman oculocerebrofacial syndrome. Journal of Medical Genetics, 2013, 50, 493-499.	1.5	40
26	From Whole Gene Deletion to Point Mutations ofEP300-Positive Rubinstein-Taybi Patients: New Insights into the Mutational Spectrum and Peculiar Clinical Hallmarks. Human Mutation, 2016, 37, 175-183.	1.1	36
27	Final Height of Patients with Turnerâ€™s Syndrome Treated with Growth Hormone (GH): Indications for GH Therapy Alone at High Doses and Late Estrogen Therapy. Journal of Clinical Endocrinology and Metabolism, 1999, 84, 4510-4515.	1.8	35
28	A mutation in PAK3 with a dual molecular effect deregulates the RAS/MAPK pathway and drives an X-linked syndromic phenotype. Human Molecular Genetics, 2014, 23, 3607-3617.	1.4	33
29	The association with Turner syndrome significantly affects the course of Hashimotoâ€™s thyroiditis in children, irrespective of karyotype. Endocrine, 2015, 50, 777-782.	1.1	33
30	Response to longâ€term growth hormone therapy in patients affected by RASopathies and growth hormone deficiency: Patterns of growth, puberty and final height data. American Journal of Medical Genetics, Part A, 2015, 167, 2786-2794.	0.7	32
31	Helicobacter pylori and Type 1 Diabetes Mellitus in Children. Journal of Pediatric Gastroenterology and Nutrition, 1999, 28, 307-309.	0.9	32
32	Epidemiology, Presentation and Long-Term Evolution of Graves' Disease in Children, Adolescents and Young Adults with Turner Syndrome. Hormone Research in Paediatrics, 2014, 81, 245-250.	0.8	31
33	A de novo nonsense mutation ofPAX6 gene in a patient with aniridia, ataxia, and mental retardation. American Journal of Medical Genetics, Part A, 2007, 143A, 1802-1805.	0.7	28
34	Barberâ€“Say syndrome and Ablepharonâ€“Macrostomia syndrome: An overview. American Journal of Medical Genetics, Part A, 2016, 170, 1989-2001.	0.7	26
35	Turner Syndrome, Insulin Sensitivity and Growth Hormone Treatment. Hormone Research in Paediatrics, 2005, 64, 51-57.	0.8	23
36	Early treatment with GH alone in Turner syndrome: prepubertal catch-up growth and waning effect. European Journal of Endocrinology, 2004, 151, 567-572.	1.9	21

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37	A syndromic form of Pierre Robin sequence is caused by 5q23 deletions encompassing FBN2 and PHAX. European Journal of Medical Genetics, 2014, 57, 587-595.	0.7	18
38	Stories of experiences of care for growth hormone deficiency: the CRESCERE project. Future Science OA, 2016, 2, FSO82.	0.9	17
39	Variants of SOS2 are a rare cause of Noonan syndrome with particular predisposition for lymphatic complications. European Journal of Human Genetics, 2021, 29, 51-60.	1.4	17
40	Adult height in girls with Turner syndrome treated from before 6 years of age with a fixed per kilogram GH dose. European Journal of Endocrinology, 2013, 169, 439-443.	1.9	16
41	Impaired GH Secretion in Patients with SHOX Deficiency and Efficacy of Recombinant Human GH Therapy. Hormone Research in Paediatrics, 2012, 78, 279-287.	0.8	15
42	The Influence of Growth Hormone Treatment on Glucose Homeostasis in GrowthHormone-Deficient Children: A Six-Year Follow-Up Study. Hormone Research in Paediatrics, 2016, 86, 196-200.	0.8	15
43	GH, ACTH, TSH, LH, and FSH Reserve in Prepubertal Girls with Congenital Adrenal Hyperplasia. Journal of Clinical Endocrinology and Metabolism, 1976, 43, 1146-1152.	1.8	14
44	Developmental Syndromes: Growth Hormone Deficiency and Treatment. Endocrine Development, 2009, 14, 114-134.	1.3	14
45	GH Therapy and first final height data in Noonanâ€like syndrome with loose anagen hair (Mazzanti) Tj ETQq1 1 0.784314 rgBT /Overlock	0.7	14
46	The Evolution of Thyroid Function after Presenting with Hashimoto Thyroiditis Is Different between Initially Euthyroid Girls with and Those without Turner Syndrome. Hormone Research in Paediatrics, 2016, 86, 403-409.	0.8	14
47	The influence of GH treatment on glucose homeostasis in girls with Turner Syndrome: a 7 years study. Journal of Clinical Endocrinology and Metabolism, 2017, 102, jc.2016-3179.	1.8	13
48	Wolfâ€“Hirschhorn syndrome-associated chromosome changes are not mediated by olfactory receptor gene clusters nor by inversion polymorphism on 4p16. Human Genetics, 2007, 122, 423-430.	1.8	12
49	Transcriptional hallmarks of noonan syndrome and noonanâ€like syndrome with loose anagen hair. Human Mutation, 2012, 33, 703-709.	1.1	12
50	Estradiol matrix patches for pubertal induction: stability of cut pieces at different temperatures. Endocrine Connections, 2019, 8, 360-366.	0.8	12
51	Correlations of phenotype and genotype in relation to morphologic remodelling of the aortic root in patients with Turnerâ€™s syndrome. Cardiology in the Young, 2009, 19, 264.	0.4	11
52	Challenges in the clinical interpretation of small de novo copy number variants in neurodevelopmental disorders. Gene, 2019, 706, 162-171.	1.0	9
53	The empowerment of translational research: lessons from laminopathies. Orphanet Journal of Rare Diseases, 2012, 7, 37.	1.2	7
54	Barber-Say Syndrome and Ablepharon-Macrostomia Syndrome: A Patient's View. Molecular Syndromology, 2017, 8, 172-178.	0.3	6

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55	Neuropsychiatric phenotype in a child with pseudohypoparathyroidism. Journal of Pediatric Neurosciences, 2016, 11, 267.	0.2	6
56	Growth in Children With Noonan Syndrome and Effects of Growth Hormone Treatment on Adult Height. Frontiers in Endocrinology, 2021, 12, 761171.	1.5	6
57	Involvement of the skull base and vault in chronic idiopathic hyperphosphatasia. Pediatric Radiology, 1999, 29, 16-18.	1.1	5
58	Generation of the Rubinstein-Taybi syndrome type 2 patient-derived induced pluripotent stem cell line (IAli001-A) carrying the EP300 exon 23 stop mutation c.3829Aâ€¯>â€¯T, p.(Lys1277*). Stem Cell Research, 2018, 30, 175-179.	0.3	4
59	<i>HDAC8</i> Loss of Function and <i>SHOX</i> Haploinsufficiency: Two Independent Genetic Defects Responsible for a Complex Phenotype. Cytogenetic and Genome Research, 2019, 157, 135-140.	0.6	3
60	Chondrodysplasia and growth failure in children after early hematopoietic stem cell transplantation for nonâ€oncologic disorders. American Journal of Medical Genetics, Part A, 2021, 185, 517-527.	0.7	3
61	Turner syndrome strategies to improve care outcomes--cardiac evaluation using new imaging techniques. Pediatric Endocrinology Reviews, 2012, 9 Suppl 2, 701-9.	1.2	2
62	Hearing Growth Defects in Turner Syndrome. , 2012, , 1437-1444.		0