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	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
2	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
3	Growth in Children With Noonan Syndrome and Effects of Growth Hormone Treatment on Adult Height. Frontiers in Endocrinology, 2021, 12, 761171.	1.5	6
4	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	2.6	48
5	Challenges in the clinical interpretation of small de novo copy number variants in neurodevelopmental disorders. Gene, 2019, 706, 162-171.	1.0	9
6	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
7	<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
8	Estradiol matrix patches for pubertal induction: stability of cut pieces at different temperatures. Endocrine Connections, 2019, 8, 360-366.	0.8	12
9	Characteristics of a nationwide cohort of patients presenting with isolated hypogonadotropic hypogonadism (IHH). European Journal of Endocrinology, 2018, 178, 23-32.	1.9	84
10	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
11	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
12	Barber-Say Syndrome and Ablepharon-Macrostomia Syndrome: A Patient's View. Molecular Syndromology, 2017, 8, 172-178.	0.3	6
13	Stories of experiences of care for growth hormone deficiency: the CRESCERE project. Future Science OA, 2016, 2, FSO82.	0.9	17
14	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
15	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
16	Barberâ€“Say syndrome and Ablepharonâ€“Macrostomia syndrome: An overview. American Journal of Medical Genetics, Part A, 2016, 170, 1989-2001.	0.7	26
17	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
18	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

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19	Neuropsychiatric phenotype in a child with pseudohypoparathyroidism. Journal of Pediatric Neurosciences, 2016, 11, 267.	0.2	6
20	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
21	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
22	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
23	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
24	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
25	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
26	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
27	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
28	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
29	Epilepsy in Mowatâ€“Wilson syndrome: Delineation of the electroclinical phenotype. American Journal of Medical Genetics, Part A, 2013, 161, 273-284.	0.7	42
30	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
31	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
32	GH Therapy and first final height data in Noonanâ€like syndrome with loose anagen hair (Mazzanti) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5	0.7	14
33	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
34	The empowerment of translational research: lessons from laminopathies. Orphanet Journal of Rare Diseases, 2012, 7, 37.	1.2	7
35	Transcriptional hallmarks of noonan syndrome and noonanâ€like syndrome with loose anagen hair. Human Mutation, 2012, 33, 703-709.	1.1	12
36	Hearing Growth Defects in Turner Syndrome. , 2012, , 1437-1444.		0

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37	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
38	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
39	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
40	A restricted spectrum of NRAS mutations causes Noonan syndrome. Nature Genetics, 2010, 42, 27-29.	9.4	271
41	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
42	GermlineBRAFmutations in Noonan, LEOPARD, and cardiofaciocutaneous syndromes: Molecular diversity and associated phenotypic spectrum. Human Mutation, 2009, 30, 695-702.	1.1	251
43	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
44	Developmental Syndromes: Growth Hormone Deficiency and Treatment. Endocrine Development, 2009, 14, 114-134.	1.3	14
45	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
46	Hearing loss in Turner syndrome: Results of a multicentric study. Journal of Endocrinological Investigation, 2008, 31, 779-783.	1.8	44
47	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
48	Gain-of-function RAF1 mutations cause Noonan and LEOPARD syndromes with hypertrophic cardiomyopathy. Nature Genetics, 2007, 39, 1007-1012.	9.4	624
49	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
50	Gonadoblastoma in Turner syndrome and Y-chromosome-derived material. American Journal of Medical Genetics, Part A, 2005, 135A, 150-154.	0.7	87
51	Turner Syndrome, Insulin Sensitivity and Growth Hormone Treatment. Hormone Research in Paediatrics, 2005, 64, 51-57.	0.8	23
52	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
53	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
54	Noonan-like syndrome with loose anagen hair: A new syndrome?. American Journal of Medical Genetics Part A, 2003, 118A, 279-286.	2.4	72

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55	Prevalence and Clinical Picture of Celiac Disease in Turner Syndrome. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 5495-5498.	1.8	161
56	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
57	Involvement of the skull base and vault in chronic idiopathic hyperphosphatasia. Pediatric Radiology, 1999, 29, 16-18.	1.1	5
58	Helicobacter pylori and Type 1 Diabetes Mellitus in Children. Journal of Pediatric Gastroenterology and Nutrition, 1999, 28, 307-309.	0.9	32
59	Congenital heart disease in patients with Turnerâ€™s syndrome. Journal of Pediatrics, 1998, 133, 688-692.	0.9	233
60	Pelvic ultrasonography in patients with Turner syndrome: Age-related findings in different karyotypes. Journal of Pediatrics, 1997, 131, 135-140.	0.9	45
61	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
62	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