Angela E Lin

List of Publications by Year
in descending order

126 papers	7,100 citations	71102 41 h-index	62596 80 g-index
129 all docs	129 docs citations	129 times ranked	7366 citing authors

#	Article	IF	Citations
1	Clinical practice guidelines for the care of girls and women with Turner syndrome: proceedings from the 2016 Cincinnati International Turner Syndrome Meeting. European Journal of Endocrinology, 2017, 177, G1-G70.	3.7	771
2	Guidelines for case classification for the national birth defects prevention study. Birth Defects Research Part A: Clinical and Molecular Teratology, 2003, 67, 193-201.	1.6	501
3	Seeking causes: Classifying and evaluating congenital heart defects in etiologic studies. Birth Defects Research Part A: Clinical and Molecular Teratology, 2007, 79, 714-727.	1.6	367
4	Cardiovascular disease in neurofibromatosis 1: Report of the NF1 Cardiovascular Task Force. Genetics in Medicine, 2002, 4, 105-111.	2.4	330
5	Cardiovascular malformations and other cardiovascular abnormalities in neurofibromatosis 1. American Journal of Medical Genetics Part A, 2000, 95, 108-117.	2.4	214
6	TFAP2A Mutations Result in Branchio-Oculo-Facial Syndrome. American Journal of Human Genetics, 2008, 82, 1171-1177.	6.2	193
7	Reviewing the evidence for mycophenolate mofetil as a new teratogen: Case report and review of the literature. American Journal of Medical Genetics, Part A, 2009, 149A, 1241-1248.	1.2	187
8	Further Delineation of Aortic Dilation, Dissection, and Rupture in Patients With Turner Syndrome. Pediatrics, 1998, 102, e12-e12.	2.1	180
9	HRAS mutation analysis in Costello syndrome: Genotype and phenotype correlation. American Journal of Medical Genetics, Part A, 2006, 140A, 1-7.	1.2	164
10	Clinical and molecular aspects of the Simpson-Golabi-Behmel syndrome. American Journal of Medical Genetics Part A, 1998, 79, 279-283.	2.4	156
11	Laterality defects in the national birth defects prevention study (1998â€“2007): Birth prevalence and descriptive epidemiology. American Journal of Medical Genetics, Part A, 2014, 164, 2581-2591.	1.2	145
12	Heterotaxy: Associated conditions and hospital-based prevalence in newborns. Genetics in Medicine, 2000, 2, 157-172.	2.4	141
13	Five additional Costello syndrome patients with rhabdomyosarcoma: Proposal for a tumor screening protocol. American Journal of Medical Genetics Part A, 2002, 108, 80-87.	2.4	138
14	Clinical Approach to Genetic Cardiomyopathy in Children. Circulation, 1996, 94, 2021-2038.	1.6	138
15	Costello syndrome: a Ras/mitogen activated protein kinase pathway syndrome (rasopathy) resulting from HRAS germline mutations. Genetics in Medicine, 2012, 14, 285-292.	2.4	135
16	Adults with genetic syndromes and cardiovascular abnormalities: clinical history and management. Genetics in Medicine, 2008, 10, 469-494.	2.4	130
17	The neuroimaging findings in Sotos syndrome. American Journal of Medical Genetics Part A, 1997, 68, 462-465.	2.4	125
18	Baraitserâ€“Winter cerebrofrontofacial syndrome: delineation of the spectrum in 42 cases. European Journal of Human Genetics, 2015, 23, 292-301.	2.8	115

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19	Further delineation of cardiac abnormalities in Costello syndrome. American Journal of Medical Genetics Part A, 2002, 111, 115-129.	2.4	104
20	Turner Syndrome Is an Independent Risk Factor for Aortic Dilation in the Young. Pediatrics, 2008, 121, e1622-e1627.	2.1	104
21	Clinical, pathological, and molecular analyses of cardiovascular abnormalities in Costello syndrome: A Ras/MAPK pathway syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 486-507.	1.2	99
22	Infants with Bochdalek diaphragmatic hernia: Sibling precurrence and monozygotic twin discordance in a hospitalâ€based malformation surveillance program. American Journal of Medical Genetics, Part A, 2005, 138A, 81-88.	1.2	87
23	Further delineation of the branchio-oculo-facial syndrome. American Journal of Medical Genetics Part A, 1995, 56, 42-59.	2.4	86
24	Delayed Diagnosis of Critical Congenital Heart Defects: Trends and Associated Factors. Pediatrics, 2014, 134, e373-e381.	2.1	85
25	Further delineation of the phenotype resulting fromBRAForMEK1germline mutations helps differentiate cardio-facio-cutaneous syndrome from Costello syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 1472-1480.	1.2	79
26	Congenital diaphragmatic hernia and associated cardiovascular malformations: Type, frequency, and impact on management. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2007, 145C, 201-216.	1.6	79
27	Cardiovascular malformations in Smith-Lemli-Opitz syndrome. , 1997, 68, 270-278.		73
28	Recurrent De Novo Mutations Affecting Residue Arg138 of Pyrroline-5-Carboxylate Synthase Cause a Progeroid Form of Autosomal-Dominant Cutis Laxa. American Journal of Human Genetics, 2015, 97, 483-492.	6.2	70
29	Costello syndrome: Clinical phenotype, genotype, and management guidelines. American Journal of Medical Genetics, Part A, 2019, 179, 1725-1744.	1.2	70
30	Genetic aspects of atrioventricular septal defects. American Journal of Medical Genetics Part A, 2000, 97, 289-296.	2.4	69
31	Clonazepam use in pregnancy and the risk of malformations. Birth Defects Research Part A: Clinical and Molecular Teratology, 2004, 70, 534-536.	1.6	66
32	Maleâ€toâ€male transmission of Costello syndrome: G12S HRAS germline mutation inherited from a father with somatic mosaicism. American Journal of Medical Genetics, Part A, 2009, 149A, 315-321.	1.2	62
33	Cardiac anomalies in the Simpson-Golabi-Behmel syndrome. American Journal of Medical Genetics Part A, 1999, 83, 378-381.	2.4	61
34	Genotypeâ€“phenotype analysis of the branchioâ€oculoâ€facial syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 22-32.	1.2	61
35	Phenotypic analysis of individuals with Costello syndrome due to HRAS p.G13C. , 2011, 155, 706-716.		55
36	Gainâ€ofâ€function mutations in SMAD4 cause a distinctive repertoire of cardiovascular phenotypes in patients with Myhre syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2617-2631.	1.2	53

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37	Autosomal and X chromosome structural variants are associated with congenital heart defects in Turner syndrome: The NHLBI GenTAC registry. American Journal of Medical Genetics, Part A, 2016, 170, 3157-3164.	1.2	53
38	Prenatal features of Costello syndrome: ultrasonographic findings and atrial tachycardia. Prenatal Diagnosis, 2009, 29, 682-690.	2.3	52
39	Sensenbrenner syndrome (Cranioectodermal dysplasia): Clinical and molecular analyses of 39 patients including two new patients. American Journal of Medical Genetics, Part A, 2013, 161, 2762-2776.	1.2	52
40	Thoracic aortic disease in two patients with juvenile polyposis syndrome and SMAD4 mutations. American Journal of Medical Genetics, Part A, 2013, 161, 185-191.	1.2	51
41	Maternal Antihypertensive Medication Use and Congenital Heart Defects. Hypertension, 2017, 69, 798-805.	2.7	51
42	Adams-Oliver syndrome associated with cardiovascular malformations. Clinical Dysmorphology, 1998, 7, 235-241.	0.3	47
43	Heterogeneity of mutational mechanisms and modes of inheritance in auriculocondylar syndrome. Journal of Medical Genetics, 2013, 50, 174-186.	3.2	44
44	An additional patient with mycophenolate mofetil embryopathy: Cardiac and facial analyses. , 2011, 155, 748-756.		43
45	Reciprocal deletion and duplication at 2q23.1 indicates a role for MBD5 in autism spectrum disorder. European Journal of Human Genetics, 2014, 22, 57-63.	2.8	42
46	Causes of Congenital Malformations. Birth Defects Research, 2018, 110, 87-91.	1.5	42
47	Expanding the clinical and molecular findings in RASA1 capillary malformation-arteriovenous malformation. European Journal of Human Genetics, 2018, 26, 1521-1536.	2.8	42
48	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
49	Cardiovascular malformations: Changes in prevalence and birth status, 1972-1990. , 1999, 84, 102-110.		41
50	Turner syndrome. Current Opinion in Endocrinology, Diabetes and Obesity, 2015, 22, 65-72.	2.3	41
51	Myhre and LAPS syndromes: clinical and molecular review of 32 patients. European Journal of Human Genetics, 2014, 22, 1272-1277.	2.8	38
52	Cardiac phenotypes in chromosome 4qâ^' syndrome with and without a deletion of the dHAND gene. Genetics in Medicine, 2002, 4, 464-467.	2.4	37
53	Cardiovascular malformations in Fryns syndrome: Is there a pathogenic role for neural crest cells?. American Journal of Medical Genetics, Part A, 2005, 139A, 186-193.	1.2	36
54	Congenital heart disease in supernumerary der(22), t(11;22) syndrome. Clinical Genetics, 1986, 29, 269-275.	2.0	35

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55	Recognition and management of adults with Turner syndrome: From the transition of adolescence through the senior years. American Journal of Medical Genetics, Part A, 2019, 179, 1987-2033.	1.2	33
56	Myocardial storage of chondroitin sulfate-containing moieties in Costello syndrome patients with severe hypertrophic cardiomyopathy. , 2005, 133A, 1-12.		32
57	Populationâ€based analysis of leftâ€and rightâ€sided diaphragmatic hernias demonstrates different frequencies of selected additional anomalies. American Journal of Medical Genetics, Part A, 2007, 143A, 3127-3136.	1.2	31
58	The diagnosis of Costello syndrome: Nomenclature in Ras/MAPK pathway disorders. American Journal of Medical Genetics, Part A, 2008, 146A, 1218-1220.	1.2	29
59	Familial Brachmann-de Lange syndrome: Further evidence for autosomal dominant inheritance and review of the literature. American Journal of Medical Genetics Part A, 1993, 47, 1064-1067.	2.4	24
60	Damaging de novo missense variants inEEF1A2lead to a developmental and degenerative epilepticâ€dyskinetic encephalopathy. Human Mutation, 2020, 41, 1263-1279.	2.5	24
61	Toward a genetic etiology of CHARGE syndrome: I. A systematic scan for submicroscopic deletions. American Journal of Medical Genetics Part A, 2003, 118A, 260-266.	2.4	23
62	Cancer occurrence in Turner syndrome and the effect of sex hormone substitution therapy. European Journal of Endocrinology, 2021, 184, 79-88.	3.7	23
63	47,XXX associated with malformations. American Journal of Medical Genetics Part A, 2004, 125A, 108-111.	2.4	21
64	Nonreentrant atrial tachycardia occurs independently of hypertrophic cardiomyopathy in RASopathy patients. American Journal of Medical Genetics, Part A, 2018, 176, 1711-1722.	1.2	21
65	Cervical artery dissection expands the cardiovascular phenotype in FBN1â€related Weillâ€“Marchesani syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 2551-2556.	1.2	20
66	Exclusion of the branchio-oto-renal syndrome locus (EYA1) from patients with branchio-oculo-facial syndrome. , 2000, 91, 387-390.		19
67	Turner syndrome and meningioma: Support for a possible increased risk of neoplasia in Turner syndrome. European Journal of Medical Genetics, 2014, 57, 269-274.	1.3	19
68	Congenital heart defects associated with aneuploidy syndromes: New insights into familiar associations. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 53-63.	1.6	19
69	High diagnostic yield in skeletal ciliopathies using massively parallel genome sequencing, structural variant screening and RNA analyses. Journal of Human Genetics, 2021, 66, 995-1008.	2.3	19
70	The Branchio-Oculo-Facial Syndrome. Cleft Palate-Craniofacial Journal, 1991, 28, 96-102.	0.9	18
71	Genetic epidemiology of cardiovascular malformations. Progress in Pediatric Cardiology, 2005, 20, 113-126.	0.4	18
72	The Active Malformations Surveillance Program, Boston in 1972â€“2012: Methodology and demographic characteristics. Birth Defects Research, 2018, 110, 148-156.	1.5	18

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73	Focus on the Heart and Aorta in Turner Syndrome. Journal of Pediatrics, 2007, 150, 572-574.	1.8	15
74	Case 4-2017. New England Journal of Medicine, 2017, 376, 562-574.	27.0	15
75	Dual diagnoses in 152 patients with Turner syndrome: Knowledge of the second condition may lead to modification of treatment and/or surveillance. American Journal of Medical Genetics, Part A, 2018, 176, 2435-2445.	1.2	15
76	Clarification of previously reported Costello syndrome patients. American Journal of Medical Genetics, Part A, 2008, 146A, 940-943.	1.2	13
77	Lesser Forms of Cleft Lip Associated With the Branchio-Oculo-Facial Syndrome. Journal of Craniofacial Surgery, 2009, 20, 608-611.	0.7	13
78	Genome sequencing reveals a deep intronic splicing ACVRL1 mutation hotspot in Hereditary Haemorrhagic Telangiectasia. Journal of Medical Genetics, 2018, 55, 824-830.	3.2	13
79	Fryns syndrome with Hirschsprung disease: Support for possible neural crest involvement. American Journal of Medical Genetics, Part A, 2005, 132A, 226-230.	1.2	12
80	Living with Costello syndrome: Quality of life issues in older individuals. American Journal of Medical Genetics, Part A, 2010, 152A, 84-90.	1.2	12
81	Cardiac transplantation in children with Noonan syndrome. Pediatric Transplantation, 2019, 23, e13535.	1.0	12
82	Gainâ€ofâ€function pathogenic variants in SMAD4 are associated with neoplasia in Myhre syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 328-337.	1.2	12
83	Proposal for a national registry to monitor women with Turner syndrome seeking assisted reproductive technology. Fertility and Sterility, 2016, 105, 1446-1448.	1.0	11
84	Genetic counseling for women with 45,X/46,XX mosaicism: Towards more personalized management. European Journal of Medical Genetics, 2021, 64, 104140.	1.3	11
85	Participation by clinical geneticists in genetic advocacy groups. American Journal of Medical Genetics Part A, 2003, 119A, 89-92.	2.4	10
86	Total is more than the sum of the parts: Phenotyping the heart in cardiovascular genetics clinics. American Journal of Medical Genetics Part A, 2004, 131A, 111-114.	2.4	10
87	Clinical geneticists in birth defects surveillance and epidemiology research programs: Past, present and future roles. Birth Defects Research Part A: Clinical and Molecular Teratology, 2009, 85, 69-75.	1.6	10
88	45,X mosaicism in a population-based biobank: implications for Turner syndrome. Genetics in Medicine, 2019, 21, 1882-1883.	2.4	10
89	Circumferential abdominal skin defect possibly due to umbilical cord encirclement. Teratology, 1999, 60, 258-259.	1.6	9
90	Impact of elective termination on the occurrence of severe birth defects identified in a hospitalâ€based active malformations surveillance program (1999 to 2002). Birth Defects Research Part A: Clinical and Molecular Teratology, 2016, 106, 659-666.	1.6	9

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91	Malformations Surveillance: Comparison between Findings at Birth and Age 1 Year. Birth Defects Research, 2018, 110, 142-147.	1.5	9
92	Hand it to the skin in Costello syndrome. Journal of Pediatrics, 2004, 144, 135.	1.8	7
93	Deletion of chromosome 8q22.1, a critical region for Nablus maskâ€like facial syndrome: Four additional cases support a role of genetic modifiers in the manifestation of the phenotype. American Journal of Medical Genetics, Part A, 2015, 167, 1400-1405.	1.2	7
94	Clinician reviewers in birth defects surveillance programs: Survey of the National Birth Defects Prevention Network. Birth Defects Research Part A: Clinical and Molecular Teratology, 2006, 76, 781-786.	1.6	6
95	Need for greater precision in reporting cardiovascular malformations. American Journal of Medical Genetics Part A, 1994, 51, 84-85.	2.4	5
96	Further delineation of van den Endeâ€Gupta syndrome: Genetic heterogeneity and overlap with congenital heart defects and skeletal malformations syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2136-2149.	1.2	5
97	Parenthood among individuals with Turner syndrome: results of an online survey of attitudes towards pregnancy, adoption, and surrogacy. Journal of Community Genetics, 2022, 13, 263-270.	1.2	5
98	Heart development and the genetic aspects of cardiovascular malformations. American Journal of Medical Genetics Part A, 2000, 97, 235-237.	2.4	4
99	Writing for Scientific Publication: Tips for Getting Started. Clinical Pediatrics, 2006, 45, 295-300.	0.8	4
100	Case 34-2016. New England Journal of Medicine, 2016, 375, 1879-1890.	27.0	4
101	â€œDonating our bodies to scienceâ€: A discussion about autopsy and organ donation in Turner syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 22-28.	1.6	4
102	Grebe syndrome in Vietnamese sisters: not Agent Orange. Clinical Genetics, 2001, 59, 25-27.	2.0	3
103	Exclusion of growth factor gene mutations as a common cause of Sotos syndrome. American Journal of Medical Genetics Part A, 2001, 98, 101-102.	2.4	3
104	Congenital polyvalvular disease expands the cardiac phenotype of the RASopathies. American Journal of Medical Genetics, Part A, 2021, 185, 1486-1493.	1.2	3
105	Aortic Dilatation with Bicuspid Aortic Valve. New England Journal of Medicine, 2014, 371, 683-683.	27.0	2
106	An application of data mining to identify potential risk factors for anophthalmia and microphthalmia. Paediatric and Perinatal Epidemiology, 2018, 32, 545-555.	1.7	2
107	Cardiovascular malformations: Changes in prevalence and birth status, 1972â€“1990. American Journal of Medical Genetics Part A, 1999, 84, 102-110.	2.4	2
108	Dermatologic findings in individuals with Turner syndrome: A cross-sectional study across the lifespan. Journal of the American Academy of Dermatology, 2022, 87, 476-479.	1.2	2

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109	Expanded cardiovascular phenotype of Myhre syndrome includes tetralogy of Fallot suggesting a role for <scp>SMAD4</scp> in human neural crest defects. American Journal of Medical Genetics, Part A, 2022, 188, 1384-1395.	1.2	2
110	LETTER TO THE EDITOR Re: First live birth after fertility preservation using vitrification of oocytes in a woman with mosaic Turner syndrome. Journal of Assisted Reproduction and Genetics, 2022, 39, 777-778.	2.5	2
111	VelÃ¡zquez' Dwarfs: A Profusion of Diagnoses. JAMA - Journal of the American Medical Association, 1989, 262, 349.	7.4	1
112	Response to: Toriello et al., â€œUpdate on the Torielloâ€“Carey Syndrome.â€•Further delineation of a young woman with deletion 1q42.12â€q42.2. American Journal of Medical Genetics, Part A, 2017, 173, 1988-1991.	1.2	1
113	M. Michael Cohen, Jr.: Author, diagnostician, geneticist, teacher, mentor, syndrome scholar extraordinaire (1937â€“2018). American Journal of Medical Genetics, Part A, 2018, 176, 1703-1705.	1.2	1
114	Depiction of ectrodactyly, sirenomelia and cyclopia in a figure by Hokusai. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 148-150.	1.6	1
115	The earliest depictions of a PIK3CAâ€Related Overgrowth Spectrum disorder: 17thâ€18th century prints of women with severe limb overgrowth. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 168-172.	1.6	1
116	Cardiac anomalies in the Simpsonâ€Golabiâ€Behmel syndrome. American Journal of Medical Genetics Part A, 1999, 83, 378-381.	2.4	1
117	O patient, where art thou?. American Journal of Medical Genetics Part A, 2002, 111, 334-334.	2.4	0
118	Classification of cardiovascular malformations associated with neuroblastoma. Journal of Pediatrics, 2005, 146, 439-441.	1.8	0
119	The Partnership of Medical Genetics and Oral and Maxillofacial Surgery When Evaluating Craniofacial Anomalies. Journal of Oral and Maxillofacial Surgery, 2015, 73, S13-S16.	1.2	0
120	Case 7-2017. New England Journal of Medicine, 2017, 376, 972-980.	27.0	0
121	Mary Ella Mascia Pierpont: Geneticist, scientist, mentor, friend (1945â€“2020). American Journal of Medical Genetics, Part A, 2021, 185, 319-323.	1.2	0
122	Lack of resemblance between Myhre syndrome and other â€œsegmental progeroidâ€•syndromes warrants restraint in applying this classification. GeroScience, 2021, 43, 459-461.	4.6	0
123	Klinefelter Syndrome and Turner Syndrome. Pediatrics in Review, 2021, 42, 272-274.	0.4	0
124	Syndromes and birth defects in art and antiquities: New perspectives on a familiar theme. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 107-110.	1.6	0
125	Use of vasoactive medications in pregnancy and the risk of stillbirth among birth defect cases. Birth Defects Research, 2022, , .	1.5	0
126	Combined X-linked familial exudative vitreoretinopathy and retinopathy of prematurity phenotype in an infant with mosaic turner syndrome with ring X chromosome. Ophthalmic Genetics, 2023, 44, 198-203.	1.2	0