

# Angela E Lin

## List of Publications by Year in descending order

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126  
papers

7,100  
citations

87401

40  
h-index

75989

78  
g-index

129  
all docs

129  
docs citations

129  
times ranked

7746  
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. <i>European Journal of Endocrinology</i> , 2017, 177, G1-G70.	1.9	771
2	Guidelines for case classification for the national birth defects prevention study. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2003, 67, 193-201.	1.6	501
3	Seeking causes: Classifying and evaluating congenital heart defects in etiologic studies. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2007, 79, 714-727.	1.6	367
4	Cardiovascular disease in neurofibromatosis 1: Report of the NF1 Cardiovascular Task Force. <i>Genetics in Medicine</i> , 2002, 4, 105-111.	1.1	330
5	Cardiovascular malformations and other cardiovascular abnormalities in neurofibromatosis 1. <i>American Journal of Medical Genetics Part A</i> , 2000, 95, 108-117.	2.4	214
6	TFAP2A Mutations Result in Branchio-Oculo-Facial Syndrome. <i>American Journal of Human Genetics</i> , 2008, 82, 1171-1177.	2.6	193
7	Reviewing the evidence for mycophenolate mofetil as a new teratogen: Case report and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 1241-1248.	0.7	187
8	Further Delineation of Aortic Dilation, Dissection, and Rupture in Patients With Turner Syndrome. <i>Pediatrics</i> , 1998, 102, e12-e12.	1.0	180
9	HRAS mutation analysis in Costello syndrome: Genotype and phenotype correlation. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 1-7.	0.7	164
10	Clinical and molecular aspects of the Simpson-Golabi-Behmel syndrome. , 1998, 79, 279-283.		156
11	Laterality defects in the national birth defects prevention study (1998-2007): Birth prevalence and descriptive epidemiology. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2581-2591.	0.7	145
12	Heterotaxy: Associated conditions and hospital-based prevalence in newborns. <i>Genetics in Medicine</i> , 2000, 2, 157-172.	1.1	141
13	Five additional Costello syndrome patients with rhabdomyosarcoma: Proposal for a tumor screening protocol. <i>American Journal of Medical Genetics Part A</i> , 2002, 108, 80-87.	2.4	138
14	Clinical Approach to Genetic Cardiomyopathy in Children. <i>Circulation</i> , 1996, 94, 2021-2038.	1.6	138
15	Costello syndrome: a Ras/mitogen activated protein kinase pathway syndrome (rasopathy) resulting from HRAS germline mutations. <i>Genetics in Medicine</i> , 2012, 14, 285-292.	1.1	135
16	Adults with genetic syndromes and cardiovascular abnormalities: clinical history and management. <i>Genetics in Medicine</i> , 2008, 10, 469-494.	1.1	130
17	The neuroimaging findings in Sotos syndrome. , 1997, 68, 462-465.		125
18	Baraitser- Winter cerebrofrontofacial syndrome: delineation of the spectrum in 42 cases. <i>European Journal of Human Genetics</i> , 2015, 23, 292-301.	1.4	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.	1.0	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.	0.7	99
22	Infants with Bochdalek diaphragmatic hernia: Sibling precurrence and monozygotic twin discordance in a hospital-based malformation surveillance program. American Journal of Medical Genetics, Part A, 2005, 138A, 81-88.	0.7	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.	1.0	85
25	Further delineation of the phenotype resulting from BRAF or MEK1 germline mutations helps differentiate cardio-facio-cutaneous syndrome from Costello syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 1472-1480.	0.7	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.	0.7	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.	2.6	70
29	Costello syndrome: Clinical phenotype, genotype, and management guidelines. American Journal of Medical Genetics, Part A, 2019, 179, 1725-1744.	0.7	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	Male-to-male transmission of Costello syndrome: G12S <i>HRAS</i> germline mutation inherited from a father with somatic mosaicism. American Journal of Medical Genetics, Part A, 2009, 149A, 315-321.	0.7	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	Genotype-phenotype analysis of the branchio-oculo-facial syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 22-32.	0.7	61
35	Phenotypic analysis of individuals with Costello syndrome due to HRAS p.G13C. , 2011, 155, 706-716.		55
36	Gain-of-function mutations in <i>SMAD4</i> cause a distinctive repertoire of cardiovascular phenotypes in patients with Myhre syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2617-2631.	0.7	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. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 3157-3164.	0.7	53
38	Prenatal features of Costello syndrome: ultrasonographic findings and atrial tachycardia. <i>Prenatal Diagnosis</i> , 2009, 29, 682-690.	1.1	52
39	Sensenbrenner syndrome (Cranioectodermal dysplasia): Clinical and molecular analyses of 39 patients including two new patients. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2762-2776.	0.7	52
40	Thoracic aortic disease in two patients with juvenile polyposis syndrome and <i>SMAD4</i> mutations. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 185-191.	0.7	51
41	Maternal Antihypertensive Medication Use and Congenital Heart Defects. <i>Hypertension</i> , 2017, 69, 798-805.	1.3	51
42	Adams-Oliver syndrome associated with cardiovascular malformations. <i>Clinical Dysmorphology</i> , 1998, 7, 235-241.	0.1	47
43	Heterogeneity of mutational mechanisms and modes of inheritance in auriculocondylar syndrome. <i>Journal of Medical Genetics</i> , 2013, 50, 174-186.	1.5	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. <i>European Journal of Human Genetics</i> , 2014, 22, 57-63.	1.4	42
46	Causes of Congenital Malformations. <i>Birth Defects Research</i> , 2018, 110, 87-91.	0.8	42
47	Expanding the clinical and molecular findings in RASA1 capillary malformation-arteriovenous malformation. <i>European Journal of Human Genetics</i> , 2018, 26, 1521-1536.	1.4	42
48	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor $\beta$ Signaling. <i>Biological Psychiatry</i> , 2020, 87, 100-112.	0.7	42
49	Cardiovascular malformations: Changes in prevalence and birth status, 1972-1990. , 1999, 84, 102-110.		41
50	Turner syndrome. <i>Current Opinion in Endocrinology, Diabetes and Obesity</i> , 2015, 22, 65-72.	1.2	41
51	Myhre and LAPS syndromes: clinical and molecular review of 32 patients. <i>European Journal of Human Genetics</i> , 2014, 22, 1272-1277.	1.4	38
52	Cardiac phenotypes in chromosome 4q <sup>+</sup> syndrome with and without a deletion of the dHAND gene. <i>Genetics in Medicine</i> , 2002, 4, 464-467.	1.1	37
53	Cardiovascular malformations in Fryns syndrome: Is there a pathogenic role for neural crest cells?. <i>American Journal of Medical Genetics, Part A</i> , 2005, 139A, 186-193.	0.7	36
54	Congenital heart disease in supernumerary der(22), t(11;22) syndrome. <i>Clinical Genetics</i> , 1986, 29, 269-275.	1.0	35

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55	Recognition and management of adults with Turner syndrome: From the transition of adolescence through the senior years. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1987-2033.	0.7	33
56	Myocardial storage of chondroitin sulfate-containing moieties in Costello syndrome patients with severe hypertrophic cardiomyopathy. , 2005, 133A, 1-12.		32
57	Population-based analysis of left- and right-sided diaphragmatic hernias demonstrates different frequencies of selected additional anomalies. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 3127-3136.	0.7	31
58	The diagnosis of Costello syndrome: Nomenclature in Ras/MAPK pathway disorders. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 1218-1220.	0.7	29
59	Familial Brachmann-de Lange syndrome: Further evidence for autosomal dominant inheritance and review of the literature. <i>American Journal of Medical Genetics Part A</i> , 1993, 47, 1064-1067.	2.4	24
60	Damaging de novo missense variants in <i>EEF1A2</i> lead to a developmental and degenerative epileptic-dyskinetic encephalopathy. <i>Human Mutation</i> , 2020, 41, 1263-1279.	1.1	24
61	Toward a genetic etiology of CHARGE syndrome: I. A systematic scan for submicroscopic deletions. <i>American Journal of Medical Genetics Part A</i> , 2003, 118A, 260-266.	2.4	23
62	Cancer occurrence in Turner syndrome and the effect of sex hormone substitution therapy. <i>European Journal of Endocrinology</i> , 2021, 184, 79-88.	1.9	23
63	47,XXX associated with malformations. <i>American Journal of Medical Genetics Part A</i> , 2004, 125A, 108-111.	2.4	21
64	Nonreentrant atrial tachycardia occurs independently of hypertrophic cardiomyopathy in RASopathy patients. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1711-1722.	0.7	21
65	Cervical artery dissection expands the cardiovascular phenotype in <i>FBN1</i> -related Weill-Marchesani syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2551-2556.	0.7	20
66	Exclusion of the branchio-oto-renal syndrome locus ( <i>EYA1</i> ) 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. <i>European Journal of Medical Genetics</i> , 2014, 57, 269-274.	0.7	19
68	Congenital heart defects associated with aneuploidy syndromes: New insights into familial associations. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2020, 184, 53-63.	0.7	19
69	High diagnostic yield in skeletal ciliopathies using massively parallel genome sequencing, structural variant screening and RNA analyses. <i>Journal of Human Genetics</i> , 2021, 66, 995-1008.	1.1	19
70	The Branchio-Oculo-Facial Syndrome. <i>Cleft Palate-Craniofacial Journal</i> , 1991, 28, 96-102.	0.5	18
71	Genetic epidemiology of cardiovascular malformations. <i>Progress in Pediatric Cardiology</i> , 2005, 20, 113-126.	0.2	18
72	The Active Malformations Surveillance Program, Boston in 1972-2012: Methodology and demographic characteristics. <i>Birth Defects Research</i> , 2018, 110, 148-156.	0.8	18

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73	Focus on the Heart and Aorta in Turner Syndrome. <i>Journal of Pediatrics</i> , 2007, 150, 572-574.	0.9	15
74	Case 4-2017. <i>New England Journal of Medicine</i> , 2017, 376, 562-574.	13.9	15
75	Dual diagnoses in 152 patients with Turner syndrome: Knowledge of the second condition may lead to modification of treatment and/or surveillance. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2435-2445.	0.7	15
76	Clarification of previously reported Costello syndrome patients. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 940-943.	0.7	13
77	Lesser Forms of Cleft Lip Associated With the Branchio-Oculo-Facial Syndrome. <i>Journal of Craniofacial Surgery</i> , 2009, 20, 608-611.	0.3	13
78	Genome sequencing reveals a deep intronic splicing <i>ACVRL1</i> mutation hotspot in Hereditary Haemorrhagic Telangiectasia. <i>Journal of Medical Genetics</i> , 2018, 55, 824-830.	1.5	13
79	Fryns syndrome with Hirschsprung disease: Support for possible neural crest involvement. <i>American Journal of Medical Genetics, Part A</i> , 2005, 132A, 226-230.	0.7	12
80	Living with Costello syndrome: Quality of life issues in older individuals. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 84-90.	0.7	12
81	Cardiac transplantation in children with Noonan syndrome. <i>Pediatric Transplantation</i> , 2019, 23, e13535.	0.5	12
82	Gain-of-function pathogenic variants in <i>SMAD4</i> are associated with neoplasia in Myhre syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 328-337.	0.7	12
83	Proposal for a national registry to monitor women with Turner syndrome seeking assisted reproductive technology. <i>Fertility and Sterility</i> , 2016, 105, 1446-1448.	0.5	11
84	Genetic counseling for women with 45,X/46,XX mosaicism: Towards more personalized management. <i>European Journal of Medical Genetics</i> , 2021, 64, 104140.	0.7	11
85	Participation by clinical geneticists in genetic advocacy groups. <i>American Journal of Medical Genetics Part A</i> , 2003, 119A, 89-92.	2.4	10
86	Total is more than the sum of the parts: Phenotyping the heart in cardiovascular genetics clinics. <i>American Journal of Medical Genetics Part A</i> , 2004, 131A, 111-114.	2.4	10
87	Clinical geneticists in birth defects surveillance and epidemiology research programs: Past, present and future roles. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2009, 85, 69-75.	1.6	10
88	45,X mosaicism in a population-based biobank: implications for Turner syndrome. <i>Genetics in Medicine</i> , 2019, 21, 1882-1883.	1.1	10
89	Circumferential abdominal skin defect possibly due to umbilical cord encirclement. <i>Teratology</i> , 1999, 60, 258-259.	1.8	9
90	Impact of elective termination on the occurrence of severe birth defects identified in a hospital-based active malformations surveillance program (1999 to 2002). <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 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.	0.8	9
92	Hand it to the skin in Costello syndrome. Journal of Pediatrics, 2004, 144, 135.	0.9	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.	0.7	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.	0.7	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.	0.5	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.	0.4	4
100	Case 34-2016. New England Journal of Medicine, 2016, 375, 1879-1890.	13.9	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.	0.7	4
102	Grebe syndrome in Vietnamese sisters: not Agent Orange. Clinical Genetics, 2001, 59, 25-27.	1.0	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.	0.7	3
105	Aortic Dilatation with Bicuspid Aortic Valve. New England Journal of Medicine, 2014, 371, 683-683.	13.9	2
106	An application of data mining to identify potential risk factors for anophthalmia and microphthalmia. Paediatric and Perinatal Epidemiology, 2018, 32, 545-555.	0.8	2
107	Cardiovascular malformations: Changes in prevalence and birth status, 1972â€1990. , 1999, 84, 102.		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.	0.6	2

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109	Expanded cardiovascular phenotype of Myhre syndrome includes tetralogy of Fallot suggesting a role for <i>SMAD4</i> in human neural crest defects. American Journal of Medical Genetics, Part A, 2022, 188, 1384-1395.	0.7	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.	1.2	2
111	Velázquez' Dwarfs: A Profusion of Diagnoses. JAMA - Journal of the American Medical Association, 1989, 262, 349.	3.8	1
112	Response to: Toriello et al., "Update on the Toriello-Carey Syndrome." Further delineation of a young woman with deletion 1q42.12q42.2. American Journal of Medical Genetics, Part A, 2017, 173, 1988-1991.	0.7	1
113	M. Michael Cohen, Jr.: Author, diagnostician, geneticist, teacher, mentor, syndrome scholar extraordinaire (1937-2018). American Journal of Medical Genetics, Part A, 2018, 176, 1703-1705.	0.7	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.	0.7	1
115	The earliest depictions of a PIK3CA-Related Overgrowth Spectrum disorder: 17th-18th century prints of women with severe limb overgrowth. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 168-172.	0.7	1
116	Cardiac anomalies in the Simpson-Golabi-Behmel syndrome. , 1999, 83, 378.		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.	0.9	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.	0.5	0
120	Case 7-2017. New England Journal of Medicine, 2017, 376, 972-980.	13.9	0
121	Mary Ella Mascia Pierpont: Geneticist, scientist, mentor, friend (1945-2020). American Journal of Medical Genetics, Part A, 2021, 185, 319-323.	0.7	0
122	Lack of resemblance between Myhre syndrome and other "segmental progeroid" syndromes warrants restraint in applying this classification. GeroScience, 2021, 43, 459-461.	2.1	0
123	Klinefelter Syndrome and Turner Syndrome. Pediatrics in Review, 2021, 42, 272-274.	0.2	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.	0.7	0
125	Use of vasoactive medications in pregnancy and the risk of stillbirth among birth defect cases. Birth Defects Research, 2022, , .	0.8	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.	0.5	0