

# Erik Iwarsson

## List of Publications by Year in descending order

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51  
papers

1,731  
citations

331670  
21  
h-index

289244  
40  
g-index

54  
all docs

54  
docs citations

54  
times ranked

2668  
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in amphiphysin 2 (BIN1) disrupt interaction with dynamin 2 and cause autosomal recessive centronuclear myopathy. <i>Nature Genetics</i> , 2007, 39, 1134-1139.	21.4	353
2	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. <i>Genome Medicine</i> , 2021, 13, 40.	8.2	116
3	Analysis of cell-free fetal <scp>DNA</scp> in maternal blood for detection of trisomy 21, 18 and 13 in a general pregnant population and in a high risk population – a systematic review and meta-analysis. <i>Acta Obstetrica Et Gynecologica Scandinavica</i> , 2017, 96, 7-18.	2.8	94
4	From cytogenetics to cytogenomics: whole-genome sequencing as a first-line test comprehensively captures the diverse spectrum of disease-causing genetic variation underlying intellectual disability. <i>Genome Medicine</i> , 2019, 11, 68.	8.2	88
5	Fetal Membrane Cells for Treatment of Steroid-Refractory Acute Graft-Versus-Host Disease. <i>Stem Cells</i> , 2013, 31, 592-601.	3.2	84
6	On the origin of trisomy 21 Down syndrome. <i>Molecular Cytogenetics</i> , 2008, 1, 21.	0.9	80
7	A high degree of aneuploidy in frozen-thawed human preimplantation embryos. <i>Human Genetics</i> , 1999, 104, 376.	3.8	78
8	Highly abnormal cleavage divisions in preimplantation embryos from translocation carriers. <i>Prenatal Diagnosis</i> , 2000, 20, 1038-1047.	2.3	74
9	On the origin of the maternal age effect in trisomy 21 Down syndrome: the Oocyte Mosaicism Selection model. <i>Reproduction</i> , 2010, 139, 1-9.	2.6	60
10	Identification of a duplication within the GDF9 gene and novel candidate genes for primary ovarian insufficiency (POI) by a customized high-resolution array comparative genomic hybridization platform. <i>Human Reproduction</i> , 2014, 29, 1818-1827.	0.9	54
11	Trisomy 21 Mosaicism: We May All Have a Touch of Down Syndrome. <i>Cytogenetic and Genome Research</i> , 2013, 139, 189-192.	1.1	42
12	Germinal and Somatic Trisomy 21 Mosaicism: How Common is it, What are the Implications for Individual Carriers and How Does it Come About?. <i>Current Genomics</i> , 2010, 11, 409-419.	1.6	41
13	Preimplantation genetic diagnosis of a large pericentric inversion of chromosome 5. <i>Molecular Human Reproduction</i> , 1998, 4, 719-723.	2.8	39
14	Preimplantation genetic diagnosis of DiGeorge syndrome. <i>Molecular Human Reproduction</i> , 1998, 4, 871-875.	2.8	36
15	Clinical outcome of treatment cycles using preimplantation genetic diagnosis for structural chromosomal abnormalities. <i>Prenatal Diagnosis</i> , 2001, 21, 781-787.	2.3	31
16	Application of single-needle blastomere biopsy in human preimplantation genetic diagnosis. , 1998, 18, 1381-1388.		29
17	On the paternal origin of trisomy 21 Down syndrome. <i>Molecular Cytogenetics</i> , 2010, 3, 4.	0.9	29
18	Chimerism resulting from parthenogenetic activation and dispermic fertilization. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 2277-2286.	1.2	28

#	ARTICLE	IF	CITATIONS
19	Positive Attitudes towards Non-Invasive Prenatal Testing (NIPT) in a Swedish Cohort of 1,003 Pregnant Women. PLoS ONE, 2016, 11, e0156088.	2.5	28
20	Amplification-free sequencing of cell-free DNA for prenatal non-invasive diagnosis of chromosomal aberrations. Genomics, 2015, 105, 150-158.	2.9	26
21	Fetal HLA-G mediated immune tolerance and interferon response in preeclampsia. EBioMedicine, 2020, 59, 102872.	6.1	25
22	Identification of putative pathogenic single nucleotide variants (SNVs) in genes associated with heart disease in 290 cases of stillbirth. PLoS ONE, 2019, 14, e0210017.	2.5	24
23	Mutation Screening and Array Comparative Genomic Hybridization Using a 180K Oligonucleotide Array in VACTERL Association. PLoS ONE, 2014, 9, e85313.	2.5	22
24	Molecular and Cytogenetic Analysis in Stillbirth: Results from 481 Consecutive Cases. Fetal Diagnosis and Therapy, 2014, 36, 326-332.	1.4	22
25	Expanding the Clinical Spectrum of Phenotypes Caused by Pathogenic Variants in <i>PLOD2</i> . Journal of Bone and Mineral Research, 2018, 33, 753-760.	2.8	20
26	Novel candidate genes for 46,XY gonadal dysgenesis identified by a customized 1ÂM array-CGH platform. European Journal of Medical Genetics, 2013, 56, 661-668.	1.3	17
27	Mapping uncertainty in genomics. Journal of Risk Research, 2018, 21, 117-128.	2.6	17
28	Broader phenotypic traits and widespread brain hypometabolism in spinocerebellar ataxia 27. Journal of Internal Medicine, 2020, 288, 103-115.	6.0	16
29	Preimplantation genetic diagnosis: twenty years of practice. Seminars in Fetal and Neonatal Medicine, 2011, 16, 74-80.	2.3	13
30	CBX2 gene analysis in patients with 46,XY and 46,XX gonadal disorders of sex development. Fertility and Sterility, 2013, 99, 819-826.e3.	1.0	13
31	Maternal Germinal Trisomy 21 in Down Syndrome. Journal of Clinical Medicine, 2014, 3, 167-175.	2.4	12
32	PGD for dystrophin gene deletions using fluorescence in situ hybridization. Molecular Human Reproduction, 2006, 12, 353-356.	2.8	11
33	Disomy 21 in spermatozoa and the paternal origin of trisomy 21 Down syndrome. Molecular Cytogenetics, 2015, 8, 67.	0.9	11
34	Highly abnormal cleavage divisions in preimplantation embryos from translocation carriers. Prenatal Diagnosis, 2000, 20, 1038-47.	2.3	11
35	Diagnostic yield using whole-genome sequencing and <i>in silico</i> panel of 281 genes associated with non-immune hydrops fetalis in clinical setting. Ultrasound in Obstetrics and Gynecology, 2022, 60, 487-493.	1.7	11
36	Knowledge and Attitudes Regarding Non-Invasive Prenatal Testing (NIPT) and Preferences for Risk Information among High School Students in Sweden. Journal of Genetic Counseling, 2017, 26, 447-454.	1.6	10

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37	No Mutations in the <i>PSMC3IP</i> Gene Identified in a Swedish Cohort of Women with Primary Ovarian Insufficiency. <i>Sexual Development</i> , 2014, 8, 146-150.	2.0	9
38	Fetal Calcifications Are Associated with Chromosomal Abnormalities. <i>PLoS ONE</i> , 2015, 10, e0123343.	2.5	9
39	Expanded HSN4 phenotype associated with two novel mutations in <i>NTRK1</i> . <i>Neuromuscular Disorders</i> , 2008, 18, 681-684.	0.6	8
40	Jumping translocation in a phenotypically normal male: A study of mosaicism in spermatozoa, lymphocytes, and fibroblasts. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 1706-1711.	1.2	7
41	Preimplantation genetic testing practices in the Nordic countries. <i>Acta Obstetrica Et Gynecologica Scandinavica</i> , 2020, 99, 707-715.	2.8	7
42	Detection rates and residual risk for a postnatal diagnosis of an atypical chromosome aberration following combined first-trimester screening. <i>Prenatal Diagnosis</i> , 2020, 40, 852-859.	2.3	7
43	Preimplantation genetic testing legislation and accessibility in the Nordic countries. <i>Acta Obstetrica Et Gynecologica Scandinavica</i> , 2020, 99, 716-721.	2.8	6
44	Lack of aneuploidy for chromosomes 15, 16, and 18 in placentas from small-for-gestational-age liveborn infants. <i>American Journal of Obstetrics and Gynecology</i> , 2008, 198, 231.e1-231.e7.	1.3	5
45	Fetal Mediastinal Tumor of Neuroepithelial Origin in a Case of Missed Abortion. <i>Pediatric and Developmental Pathology</i> , 2012, 15, 511-513.	1.0	3
46	Confined placental mosaicism of Duchenne muscular dystrophy: a case report. <i>Molecular Cytogenetics</i> , 2020, 13, 51.	0.9	2
47	Comment on "Origin of trisomy: no evidence to support the ovarian mosaicism theory". <i>Prenatal Diagnosis</i> , 2012, 32, 1221-1221.	2.3	1
48	C.O.1 Identification of a new gene mutated in autosomal recessive centronuclear myopathy, and functional links with the dominant form. <i>Neuromuscular Disorders</i> , 2007, 17, 833.	0.6	0
49	Fetal Membrane Cells for Treatment of Steroid-Refractory Acute Graft-Versus-Host Disease. <i>Biology of Blood and Marrow Transplantation</i> , 2013, 19, S141-S142.	2.0	0
50	Costs And Cost-Effectiveness Of Non-Invasive Prenatal Diagnosis (Nipt) For Detection Of Trisomy 21 In Sweden. <i>Value in Health</i> , 2015, 18, A352.	0.3	0
51	Preimplantation Genetic Diagnosis"An Overview. , 2004, , 1065-1069.		0