Elena V Semina

List of Publications by Year
in descending order

63 papers	3,878 citations	186265 28 h-index	128289 60 g-index
66 all docs	66 docs citations	66 times ranked	3217 citing authors

#	Article	IF	Citations
1	Review of 37 patients with <scp>SOX2</scp> pathogenic variants collected by the Anophthalmia/Microphthalmia Clinical Registry and <scp>DNA</scp> research study. American Journal of Medical Genetics, Part A, 2022, 188, 187-198.	1.2	9
2	SOX11 variants cause a neurodevelopmental disorder with infrequent ocular malformations and hypogonadotropic hypogonadism and with distinct DNA methylation profile. Genetics in Medicine, 2022, 24, 1261-1273.	2.4	14
3	Novel Genetic Diagnoses in Septo-Optic Dysplasia. Genes, 2022, 13, 1165.	2.4	3
4	Dominant variants in <scp>PRR12</scp> result in unilateral or bilateral complex microphthalmia. Clinical Genetics, 2021, 99, 437-442.	2.0	5
5	Genetic disruption of zebrafish mab21l1 reveals a conserved role in eye development and affected pathways. Developmental Dynamics, 2021, 250, 1056-1073.	1.8	8
6	Comprehensive phenotypic and functional analysis of dominant and recessiveFOXE3alleles in ocular developmental disorders. Human Molecular Genetics, 2021, 30, 1591-1606.	2.9	6
7	Identification of missense MAB21L1 variants in microphthalmia and aniridia. Human Mutation, 2021, 42, 877-890.	2.5	13
8	WDR37 syndrome: identification of a distinct new cluster of disease-associated variants and functional analyses of mutant proteins. Human Genetics, 2021, 140, 1775-1789.	3.8	4
9	Novel variants in CDH2 are associated with a new syndrome including Peters anomaly. Clinical Genetics, 2020, 97, 502-508.	2.0	13
10	Compound heterozygous splicing <scp>CDON</scp> variants result in isolated ocular coloboma. Clinical Genetics, 2020, 98, 486-492.	2.0	4
11	Disruption of foxc1 genes in zebrafish results in dosage-dependent phenotypes overlapping Axenfeld-Rieger syndrome. Human Molecular Genetics, 2020, 29, 2723-2735.	2.9	15
12	De Novo Missense Variants in WDR37 Cause a Severe Multisystemic Syndrome. American Journal of Human Genetics, 2019, 105, 425-433.	6.2	24
13	Genetic landscape of isolated pediatric cataracts: extreme heterogeneity and variable inheritance patterns within genes. Human Genetics, 2019, 138, 847-863.	3.8	33
14	PITX2 deficiency and associated human disease: insights from the zebrafish model. Human Molecular Genetics, 2018, 27, 1675-1695.	2.9	64
15	Identification and functional analysis of anADAMTSL1variant associated with a complex phenotype including congenital glaucoma, craniofacial, and other systemic features in a threeâ€generation human pedigree. Human Mutation, 2017, 38, 1485-1490.	2.5	25
16	Mutations of conserved non-coding elements of PITX2 in patients with ocular dysgenesis and developmental glaucoma. Human Molecular Genetics, 2017, 26, 3630-3638.	2.9	28
17	Functional characterization of zebrafish orthologs of the human Beta 3-Glucosyltransferase B3GLCT gene mutated in Peters Plus Syndrome. PLoS ONE, 2017, 12, e0184903.	2.5	12
18	8q21.11 microdeletion in two patients with syndromic peters anomaly. American Journal of Medical Genetics, Part A, 2016, 170, 2471-2475.	1.2	16

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19	Case report of homozygous deletion involving the first coding exons of GCNT2 isoforms A and B and part of the upstream region of TFAP2A in congenital cataract. BMC Medical Genetics, 2016, 17, 64.	2.1	8
20	Novel mutations in PAX6, OTX2 and NDP in anophthalmia, microphthalmia and coloboma. European Journal of Human Genetics, 2016, 24, 535-541.	2.8	62
21	Analysis of in pediatric and adult glaucoma and other ocular phenotypes. Molecular Vision, 2016, 22, 1229-1238.	1.1	6
22	EFTUD2 deficiency in vertebrates: Identification of a novel human mutation and generation of a zebrafish model. Birth Defects Research Part A: Clinical and Molecular Teratology, 2015, 103, 630-640.	1.6	29
23	Identification of an Aluâ€repeatâ€mediated deletion of <scp>OPTN</scp> upstream region in a patient with a complex ocular phenotype. Molecular Genetics & Genomic Medicine, 2015, 3, 490-499.	1.2	16
24	Mutations inSIPA1L3cause eye defects through disruption of cell polarity and cytoskeleton organization. Human Molecular Genetics, 2015, 24, 5789-5804.	2.9	32
25	Mutations in MAB21L2 Result in Ocular Coloboma, Microcornea and Cataracts. PLoS Genetics, 2015, 11, e1005002.	3.5	48
26	A Case of 22q11.2 Deletion Syndrome with Peters Anomaly, Congenital Glaucoma, and Heterozygous Mutation inCYP1B1. Ophthalmic Genetics, 2015, 36, 92-94.	1.2	4
27	Conserved genetic pathways associated with microphthalmia, anophthalmia, and coloboma. Birth Defects Research Part C: Embryo Today Reviews, 2015, 105, 96-113.	3.6	62
28	Lens Extrusion fromLaminin Alpha 1Mutant Zebrafish. Scientific World Journal, The, 2014, 2014, 1-9.	2.1	12
29	Novel and recurrent PITX3 mutations in Belgian families with autosomal dominant congenital cataract and anterior segment dysgenesis have similar phenotypic and functional characteristics. Orphanet Journal of Rare Diseases, 2014, 9, 26.	2.7	30
30	Whole exome sequence analysis of Peters anomaly. Human Genetics, 2014, 133, 1497-1511.	3.8	62
31	Lens-Specific Transcription Factors and Their Roles in Diagnosis and Treatment of Human Congenital Cataract. , 2014, , 105-130.		0
32	Identification of a novel C-terminal extension mutation in EPHA2 in a family affected with congenital cataract. Molecular Vision, 2014, 20, 836-42.	1.1	15
33	Whole exome sequencing in dominant cataract identifies a new causative factor, CRYBA2, and a variety of novel alleles in known genes. Human Genetics, 2013, 132, 761-770.	3.8	72
34	PITX2 and FOXC1 spectrum of mutations in ocular syndromes. European Journal of Human Genetics, 2012, 20, 1224-1233.	2.8	137
35	pitx2 Deficiency Results in Abnormal Ocular and Craniofacial Development in Zebrafish. PLoS ONE, 2012, 7, e30896.	2.5	51
36	Analysis of FOXD3 sequence variation in human ocular disease. Molecular Vision, 2012, 18, 1740-9.	1.1	29

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37	Potential Novel Mechanism for Axenfeld-Rieger Syndrome: Deletion of a Distant Region Containing Regulatory Elements ofPITX2. , 2011, 52, 1450.		63
38	MIP/Aquaporin 0 Represents a Direct Transcriptional Target of PITX3 in the Developing Lens. PLoS ONE, 2011, 6, e21122.	2.5	25
39	Genetics of anterior segment dysgenesis disorders. Current Opinion in Ophthalmology, 2011, 22, 314-324.	2.9	130
40	BMP4 loss-of-function mutations in developmental eye disorders including SHORT syndrome. Human Genetics, 2011, 130, 495-504.	3.8	92
41	VSX2 mutations in autosomal recessive microphthalmia. Molecular Vision, 2011, 17, 2527-32.	1.1	24
42	FOXE3 plays a significant role in autosomal recessive microphthalmia. American Journal of Medical Genetics, Part A, 2010, 152A, 582-590.	1.2	50
43	Examination of SOX2 in variable ocular conditions identifies a recurrent deletion in microphthalmia and lack of mutations in other phenotypes. Molecular Vision, 2010, 16, 768-73.	1.1	15
44	Identification of dominant FOXE3 and PAX6 mutations in patients with congenital cataract and aniridia. Molecular Vision, 2010, 16, 1705-11.	1.1	43
45	Novel SOX2 mutations and genotypeâ€“phenotype correlation in anophthalmia and microphthalmia. American Journal of Medical Genetics, Part A, 2009, 149A, 2706-2715.	1.2	89
46	Mutation analysis of B3GALTL in Peters Plus syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 2603-2610.	1.2	51
47	Functional analysis of human mutations in homeodomain transcription factor PITX3. BMC Molecular Biology, 2007, 8, 84.	3.0	31
48	Mutations in laminin alpha 1 result in complex, lens-independent ocular phenotypes in zebrafish. Developmental Biology, 2006, 299, 63-77.	2.0	65
49	Analysis of RNA splicing defects in PITX2 mutants supports a gene dosage model of Axenfeld-Rieger syndrome. BMC Medical Genetics, 2006, 7, 59.	2.1	20
50	An unusual class ofPITX2 mutations in Axenfeld-Rieger syndrome. Birth Defects Research Part A: Clinical and Molecular Teratology, 2006, 76, 175-181.	1.6	27
51	Current molecular understanding of Axenfeldâ€“Rieger syndrome. Expert Reviews in Molecular Medicine, 2005, 7, 1-17.	3.9	90
52	Differential Regulation of Gene Expression by PITX2 Isoforms. Journal of Biological Chemistry, 2002, 277, 25001-25010.	3.4	142
53	A molecular basis for differential developmental anomalies in Axenfeld-Rieger syndrome. Human Molecular Genetics, 2002, 11, 743-753.	2.9	60
54	Mutations in the human forkhead transcription factor FOXE3 associated with anterior segment ocular dysgenesis and cataracts. Human Molecular Genetics, 2001, 10, 231-236.	2.9	184

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55	Identification of a Dominant Negative Homeodomain Mutation in Rieger Syndrome. Journal of Biological Chemistry, 2001, 276, 23034-23041.	3.4	82
56	The Pitx2 protein in mouse development. Developmental Dynamics, 2000, 218, 195-200.	1.8	126
57	Exclusion of the branchio-oto-renal syndrome locus (EYA1) from patients with branchio-oculo-facial syndrome. , 2000, 91, 387-390.		19
58	Antagonistic Signals between BMP4 and FGF8 Define the Expression of Pitx1 and Pitx2 in Mouse Tooth-Forming Anlage. Developmental Biology, 2000, 217, 323-332.	2.0	183
59	Cloning and chromosomal localization of two novel human genes encoding LIM-domain binding factors CLIM1 and CLIM2/LDB1/NLI. Mammalian Genome, 1998, 9, 921-924.	2.2	10
60	Assignment of gene responsible for progressive pseudorheumatoid dysplasia to chromosome 6 and examination of COL10A1 as candidate gene. European Journal of Human Genetics, 1998, 6, 251-256.	2.8	22
61	A novel homeobox gene PITX3 is mutated in families with autosomal-dominant cataracts and ASMD. Nature Genetics, 1998, 19, 167-170.	21.4	371
62	A new human homeobox gene OGI2X is a member of the most conserved homeobox gene family and is expressed during heart development in mouse. Human Molecular Genetics, 1998, 7, 415-422.	2.9	40
63	Cloning and characterization of a novel bicoid-related homeobox transcription factor gene, RIEG, involved in Rieger syndrome. Nature Genetics, 1996, 14, 392-399.	21.4	852