

Christina Gerth-Kahlert

List of Publications by Year in descending order

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Version: 2024-02-01

45
papers

588
citations

840776

11
h-index

713466

21
g-index

50
all docs

50
docs citations

50
times ranked

1166
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical and mutation analysis of 51 probands with anophthalmia and/or severe microphthalmia from a single center. <i>Molecular Genetics & Genomic Medicine</i> , 2013, 1, 15-31.	1.2	79
2	Next generation sequencing based identification of disease-associated mutations in Swiss patients with retinal dystrophies. <i>Scientific Reports</i> , 2016, 6, 28755.	3.3	62
3	A secreted WNT-ligand-binding domain of FZD5 generated by a frameshift mutation causes autosomal dominant coloboma. <i>Human Molecular Genetics</i> , 2016, 25, 1382-1391.	2.9	40
4	Incidence of retinopathy of prematurity (ROP) and ROP treatment in Switzerland 2006â€“2015: a population-based analysis. <i>Archives of Disease in Childhood: Fetal and Neonatal Edition</i> , 2018, 103, F337-F342.	2.8	38
5	Biallelic Mutations in <i>CRB1</i> Underlie Autosomal Recessive Familial Foveal Retinoschisis. , 2016, 57, 2637.		34
6	Optical Coherence Tomography and Magnetic Resonance Imaging in Multiple Sclerosis and Neuromyelitis Optica Spectrum Disorder. <i>International Journal of Molecular Sciences</i> , 2016, 17, 1894.	4.1	28
7	Dental Phenotype in Jalili Syndrome Due to a c.1312 dupC Homozygous Mutation in the <i>CNNM4</i> Gene. <i>PLoS ONE</i> , 2013, 8, e78529.	2.5	27
8	Outer Retinal Dysfunction in the Absence of Structural Abnormalities in Multiple Sclerosis. , 2018, 59, 549.		26
9	Retinal Ganglion Cell Topography in Patients With Visual Pathway Pathology. <i>Journal of Neuro-Ophthalmology</i> , 2018, 38, 172-178.	0.8	20
10	Genetic Analysis in a Swiss Cohort of Bilateral Congenital Cataract. <i>JAMA Ophthalmology</i> , 2021, 139, 691.	2.5	18
11	Higher incidence of retinopathy of prematurity in extremely preterm infants associated with improved survival rates. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2020, 109, 2033-2039.	1.5	17
12	Whole Exome Sequencing in Coloboma/Microphthalmia: Identification of Novel and Recurrent Variants in Seven Genes. <i>Genes</i> , 2021, 12, 65.	2.4	16
13	Exome Sequencing in a Swiss Childhood Glaucoma Cohort Reveals <i>CYP1B1</i> and <i>FOXC1</i> Variants as Most Frequent Causes. <i>Translational Vision Science and Technology</i> , 2020, 9, 47.	2.2	15
14	<i>C2orf71</i> Mutations as a Frequent Cause of Autosomal-Recessive Retinitis Pigmentosa: Clinical Analysis and Presentation of 8 Novel Mutations. , 2017, 58, 3840.		13
15	Genotypeâ€“phenotype spectrum in isolated and syndromic nanophthalmos. <i>Acta Ophthalmologica</i> , 2021, 99, e594-e607.	1.1	13
16	Clinical utility gene card for: Non-Syndromic Microphthalmia Including Next-Generation Sequencing-Based Approaches. <i>European Journal of Human Genetics</i> , 2017, 25, 512-512.	2.8	12
17	Confirmation of Ogden syndrome as an X-linked recessive fatal disorder due to a recurrent <i>NAA10</i> variant and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2546-2560.	1.2	12
18	Maculopathy following exposure to visible and infrared radiation from a laser pointer: a clinical case study. <i>Documenta Ophthalmologica</i> , 2016, 132, 147-155.	2.2	11

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19	Atonal homolog 7 (ATOH7) loss-of-function mutations in predominant bilateral optic nerve hypoplasia. <i>Human Molecular Genetics</i> , 2019, 29, 132-148.	2.9	11
20	Infantile hemangiomas with conjunctival involvement: An underreported occurrence. <i>Pediatric Dermatology</i> , 2017, 34, 681-685.	0.9	9
21	Outcome of Pediatric Cataract Surgeries in a Tertiary Center in Switzerland. <i>Journal of Ophthalmology</i> , 2018, 2018, 1-10.	1.3	9
22	Bilateral vitreous hemorrhage in a newborn with Stickler syndrome associated with a novel COL2A1 mutation. <i>Journal of AAPOS</i> , 2011, 15, 311-313.	0.3	8
23	Long-Range PCR-Based NGS Applications to Diagnose Mendelian Retinal Diseases. <i>International Journal of Molecular Sciences</i> , 2021, 22, 1508.	4.1	8
24	Prediction of ROP Treatment and Evaluation of Screening Criteria in VLBW Infants—a Population Based Analysis. <i>Pediatric Research</i> , 2018, 84, 632-638.	2.3	7
25	Genotype-Phenotype Analysis of a Novel Recessive and a Recurrent Dominant <i>SNRNP200</i> Variant Causing Retinitis Pigmentosa. , 2019, 60, 2822.		7
26	Characterization of two novel intronic OPA1 mutations resulting in aberrant pre-mRNA splicing. <i>BMC Medical Genetics</i> , 2017, 18, 22.	2.1	5
27	Multisystem involvement, defective lysosomes and impaired autophagy in a novel rat model of nephropathic cystinosis. <i>Human Molecular Genetics</i> , 2022, 31, 2262-2278.	2.9	5
28	Horizontal Gaze Palsy in Two Brothers with Compound Heterozygous ROBO3 Gene Mutations. <i>Neuropediatrics</i> , 2017, 48, 057-058.	0.6	4
29	Colour vision testing in young children with reduced visual acuity. <i>Acta Ophthalmologica</i> , 2020, 98, e113-e120.	1.1	4
30	Long-Term Follow-Up in Children with Anisocoria: Cocaine Test Results and Patient Outcome. <i>Journal of Ophthalmology</i> , 2017, 2017, 1-6.	1.3	3
31	Absence of Goniodysgenesis in Patients with Chromosome 13Q Microdeletion-Related Microcoria. <i>Ophthalmology Glaucoma</i> , 2018, 1, 145-147.	1.9	3
32	Optic Disc Doubling or Pseudo-Optic Disc in Colobomatous Retinal Abnormality?. <i>Journal of Neuro-Ophthalmology</i> , 2013, 33, 412.	0.8	2
33	Congenital Lamellar Cataract. <i>JAMA Ophthalmology</i> , 2014, 132, 1122.	2.5	2
34	Senescent Changes and Topography of the Dark-Adapted Multifocal Electroretinogram. , 2017, 58, 1323.		2
35	Spontaneous Nystagmus in the Dark in an Infantile Nystagmus Patient May Represent Negative Optokinetic Afternystagmus. <i>Frontiers in Neurology</i> , 2018, 9, 151.	2.4	2
36	Homozygosity for a Novel DOCK7 Variant Due to Segmental Uniparental Isodisomy of Chromosome 1 Associated with Early Infantile Epileptic Encephalopathy (EIEE) and Cortical Visual Impairment. <i>International Journal of Molecular Sciences</i> , 2022, 23, 7382.	4.1	2

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37	Alternating IV Nerve Palsy and Ptosis as a First Sign of Childhood Ocular Myasthenia Gravis. <i>Pediatric Neurology</i> , 2015, 52, 460-461.	2.1	1
38	Iris cyst in a child with Aicardi syndrome: a novel association. <i>Journal of AAPOS</i> , 2016, 20, 451-452.	0.3	1
39	Teaching Neuro <i>Images</i> : Recurrent oculomotor palsies caused by neurosarcoidosis. <i>Neurology</i> , 2016, 87, e31-2.	1.1	1
40	Unusual retinopathy in a child with severe combined immune deficiency. <i>Ophthalmic Genetics</i> , 2018, 39, 92-94.	1.2	1
41	Commentary: Outer Retinal Dysfunction on Multifocal Electroretinography May Help Differentiating Multiple Sclerosis From Neuromyelitis Optica Spectrum Disorder. <i>Frontiers in Neurology</i> , 2020, 11, 282.	2.4	1
42	Challenges in Patients with Trisomy 21: A Review of Current Knowledge and Recommendations. <i>Journal of Ophthalmology</i> , 2021, 2021, 1-7.	1.3	1
43	A three-year longitudinal study of retinal function and structure in patients with multiple sclerosis. <i>Documenta Ophthalmologica</i> , 2022, 144, 3-16.	2.2	1
44	Cocaine test results in children with anisocoria. <i>Journal of AAPOS</i> , 2016, 20, e31.	0.3	0
45	Discriminative validity of color vision tests in young children: comparison of normal and impaired vision subjects. <i>Journal of AAPOS</i> , 2018, 22, e61.	0.3	0