Christina Gerth-Kahlert

List of Publications by Year
in descending order

45	588	840776 11	713466 21
papers	citations	h-index	g-index

50	50	50	1166
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A three-year longitudinal study of retinal function and structure in patients with multiple sclerosis. Documenta Ophthalmologica, 2022, 144, 3-16.	2.2	1
2	Multisystem involvement, defective lysosomes and impaired autophagy in a novel rat model of nephropathic cystinosis. Human Molecular Genetics, 2022, 31, 2262-2278.	2.9	5
3	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. International Journal of Molecular Sciences, 2022, 23, 7382.	4.1	2
4	Genotypeâ€“phenotype spectrum in isolated and syndromic nanophthalmos. Acta Ophthalmologica, 2021, 99, e594-e607.	1.1	13
5	Whole Exome Sequencing in Coloboma/Microphthalmia: Identification of Novel and Recurrent Variants in Seven Genes. Genes, 2021, 12, 65.	2.4	16
6	Long-Range PCR-Based NGS Applications to Diagnose Mendelian Retinal Diseases. International Journal of Molecular Sciences, 2021, 22, 1508.	4.1	8
7	Challenges in Patients with Trisomy 21: A Review of Current Knowledge and Recommendations. Journal of Ophthalmology, 2021, 2021, 1-7.	1.3	1
8	Confirmation of Ogden syndrome as an Xâ€linked recessive fatal disorder due to a recurrent <scp>NAA10</scp> variant and review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 2546-2560.	1.2	12
9	Genetic Analysis in a Swiss Cohort of Bilateral Congenital Cataract. JAMA Ophthalmology, 2021, 139, 691.	2.5	18
10	Colour vision testing in young children with reduced visual acuity. Acta Ophthalmologica, 2020, 98, e113-e120.	1.1	4
11	Exome Sequencing in a Swiss Childhood Glaucoma Cohort Reveals CYP1B1 and FOXC1 Variants as Most Frequent Causes. Translational Vision Science and Technology, 2020, 9, 47.	2.2	15
12	Commentary: Outer Retinal Dysfunction on Multifocal Electroretinography May Help Differentiating Multiple Sclerosis From Neuromyelitis Optica Spectrum Disorder. Frontiers in Neurology, 2020, 11, 282.	2.4	1
13	Higher incidence of retinopathy of prematurity in extremely preterm infants associated with improved survival rates. Acta Paediatrica, International Journal of Paediatrics, 2020, 109, 2033-2039.	1.5	17
14	Genotypeâ€“Phenotype Analysis of a Novel Recessive and a Recurrent Dominant SNRNP200 Variant Causing Retinitis Pigmentosa. , 2019, 60, 2822.		7
15	Atonal homolog 7 (ATOH7) loss-of-function mutations in predominant bilateral optic nerve hypoplasia. Human Molecular Genetics, 2019, 29, 132-148.	2.9	11
16	Retinal Ganglion Cell Topography in Patients With Visual Pathway Pathology. Journal of Neuro-Ophthalmology, 2018, 38, 172-178.	0.8	20
17	Incidence of retinopathy of prematurity (ROP) and ROP treatment in Switzerland 2006â€“2015: a population-based analysis. Archives of Disease in Childhood: Fetal and Neonatal Edition, 2018, 103, F337-F342.	2.8	38
18	Unusual retinopathy in a child with severe combined immune deficiency. Ophthalmic Genetics, 2018, 39, 92-94.	1.2	1

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19	Absence of Goniodysgenesis in Patients with Chromosome 13Q Microdeletion-Related Microcoria. Ophthalmology Glaucoma, 2018, 1, 145-147.	1.9	3
20	Discriminative validity of color vision tests in young children: comparison of normal and impaired vision subjects. Journal of AAPOS, 2018, 22, e61.	0.3	0
21	Prediction of ROP Treatment and Evaluation of Screening Criteria in VLBW Infantsâ€“a Population Based Analysis. Pediatric Research, 2018, 84, 632-638.	2.3	7
22	Outcome of Pediatric Cataract Surgeries in a Tertiary Center in Switzerland. Journal of Ophthalmology, 2018, 2018, 1-10.	1.3	9
23	Spontaneous Nystagmus in the Dark in an Infantile Nystagmus Patient May Represent Negative Optokinetic Afternystagmus. Frontiers in Neurology, 2018, 9, 151.	2.4	2
24	Outer Retinal Dysfunction in the Absence of Structural Abnormalities in Multiple Sclerosis. , 2018, 59, 549.		26
25	Clinical utility gene card for: Non-Syndromic Microphthalmia Including Next-Generation Sequencing-Based Approaches. European Journal of Human Genetics, 2017, 25, 512-512.	2.8	12
26	Horizontal Gaze Palsy in Two Brothers with Compound Heterozygous ROBO3 Gene Mutations. Neuropediatrics, 2017, 48, 057-058.	0.6	4
27	Infantile hemangiomas with conjunctival involvement: AnÂunderreported occurrence. Pediatric Dermatology, 2017, 34, 681-685.	0.9	9
28	Characterization of two novel intronic OPA1 mutations resulting in aberrant pre-mRNA splicing. BMC Medical Genetics, 2017, 18, 22.	2.1	5
29	C2orf71 Mutations as a Frequent Cause of Autosomal-Recessive Retinitis Pigmentosa: Clinical Analysis and Presentation of 8 Novel Mutations. , 2017, 58, 3840.		13
30	Long-Term Follow-Up in Children with Anisocoria: Cocaine Test Results and Patient Outcome. Journal of Ophthalmology, 2017, 2017, 1-6.	1.3	3
31	Senescent Changes and Topography of the Dark-Adapted Multifocal Electroretinogram. , 2017, 58, 1323.		2
32	Biallelic Mutations inCRB1Underlie Autosomal Recessive Familial Foveal Retinoschisis. , 2016, 57, 2637.		34
33	Optical Coherence Tomography and Magnetic Resonance Imaging in Multiple Sclerosis and Neuromyelitis Optica Spectrum Disorder. International Journal of Molecular Sciences, 2016, 17, 1894.	4.1	28
34	Iris cyst in a child with Aicardi syndrome: a novel association. Journal of AAPOS, 2016, 20, 451-452.	0.3	1
35	Maculopathy following exposure to visible and infrared radiation from a laser pointer: a clinical case study. Documenta Ophthalmologica, 2016, 132, 147-155.	2.2	11
36	Cocaine test results in children with anisocoria. Journal of AAPOS, 2016, 20, e31.	0.3	0

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37	Teaching Neuro Images : Recurrent oculomotor palsies caused by neurosarcoidosis. Neurology, 2016, 87, e31-2.	1.1	1
38	Next generation sequencing based identification of disease-associated mutations in Swiss patients with retinal dystrophies. Scientific Reports, 2016, 6, 28755.	3.3	62
39	A secreted WNT-ligand-binding domain of FZD5 generated by a frameshift mutation causes autosomal dominant coloboma. Human Molecular Genetics, 2016, 25, 1382-1391.	2.9	40
40	Alternating IV Nerve Palsy and Ptosis as a First Sign of Childhood Ocular Myasthenia Gravis. Pediatric Neurology, 2015, 52, 460-461.	2.1	1
41	Congenital Lamellar Cataract. JAMA Ophthalmology, 2014, 132, 1122.	2.5	2
42	Optic Disc Doubling or Pseudo-Optic Disc in Colobomatous Retinal Abnormality?. Journal of Neuro-Ophthalmology, 2013, 33, 412.	0.8	2
43	Clinical and mutation analysis of 51 probands with anophthalmia and/or severe microphthalmia from a single center. Molecular Genetics & Genomic Medicine, 2013, 1, 15-31.	1.2	79
44	Dental Phenotype in Jalili Syndrome Due to a c.1312 dupC Homozygous Mutation in the CNNM4 Gene. PLoS ONE, 2013, 8, e78529.	2.5	27
45	Bilateral vitreous hemorrhage in a newborn with Stickler syndrome associated with a novel COL2A1 mutation. Journal of AAPOS, 2011, 15, 311-313.	0.3	8