Christina Gerth-Kahlert, MD
ORCID: 0000-0001-6298-615X

Originalarbeiten

2021

A three-year longitudinal study of retinal function and structure in patients with multiple sclerosis
Hanson JVM, Ng M-Y, Hayward-Koennecke HK, Schippling S, Reeve KA, **Gerth-Kahlert C**
Documenta Ophthalmologica, accepted 15.9.2021

Challenges in Patients with Trisomy 21: A Review of Current Knowledge and Recommendations.
Robinson J, Pompe MT, **Gerth-Kahlert C.**
J Ophthalmol. 2021 May 26;2021:8870680.

Confirmation of Ogden syndrome as an X-linked recessive fatal disorder due to a recurrent NAA10 variant and review of the literature.
Gogoll L, Steindl K, Joset P, Zweier M, Baumer A, **Gerth-Kahlert C**, Tutschek B, Rauch A.
Am J Med Genet A. 2021 Aug;185(8):2546-2560.

Genetic analysis in a Swiss Cohort of Bilateral Congenital Cataract.
Rechsteiner D*, Issler L*, Koller S, Lang E, Bähr L, Feil S, Rüegger CM, Kottke R, Toelle Stucki SP, Zweifel N, Steindl K, Joset P, Zweier, M, Suter A-A, Gogoll L, Haas C, Berger W*, **Gerth-Kahlert C***.
(*authors contributed equally to this work as first/ last author)
JAMA Ophthalmol. 2021 Jul 1;139(7):691-700.

Long-range PCR-based NGS applications to diagnose Mendelian retinal diseases.
Maggi J, Koller S, Bähr L, Feil S, Kivrak Pfiffner F, Hanson JVM, Maspoli A, **Gerth-Kahlert C**, Berger W.
Int J Mol Sci, 2021 Feb 3;22(4):1508.

Akut erworbene konkomitante Esotropie Typ 2 - eine retrospektive Analyse.
Geiger HG, Simonsz-Tooth, B, **Gerth-Kahlert C.**
Klin Monatsbl. Augenheilkunde, Apr;238(4):504-509.

The 'Eyelet sign' as an MRI clue for inflammatory Brown's syndrome.
Fierz FC, Landau K, Kottke R, Wichmann W, Sturm V, Weber KP, **Gerth-Kahlert C.**
J Neuroophthalmology, 2021 Apr 14.

Whole Exome Sequencing in Coloboma/ Microphthalmia: Identification of Novel and Recurrent Variants in Seven Genes.
Haug P, Koller S, Maggi J, Lang E, Feil S, Wlodarczyk A, Bähr L, Steindl K, Rohrbach M, **Gerth-Kahlert C***, Berger W*. (*Both authors contributed equally to this work)
Genes, 2021, Jan 6;12(1):65. doi: 10.3390/genes12010065.

Genotype-phenotype spectrum in isolated and syndromic nanophthalmos.
Lang E, Koller S, Atac D, Pfäffli OA, Hanson JVM, Feil S, Bähr L, Bahr A, Kottke R, Joset P, Fasler K, Barthelmes D, Steindl K, Konrad D, Wille DA, Berger W, **Gerth-Kahlert** C.
Acta Ophthalmol. 2021 Jun;99(4):e594-e607.

2020

Exome Sequencing in a Swiss Childhood Glaucoma Cohort Reveals *CYP1B1* and *FOXC1* Variants as Most Frequent Causes.
Lang E, Koller S, Bähr L, Töteberg-Harms M, Atac D, Roulez F, Bahr A, Steindl K, Feil S, Berger W, **Gerth-Kahlert** C.
Transl Vis Sci Technol. 2020 Jun 30;9(7):47.

Higher incidence of retinopathy of prematurity in extremely preterm infants associated with improved survival rates.
Taner A, Tekle S, Hothorn T, Adams M, Bassler D, **Gerth-Kahlert** C..
Acta Paediatr. 2020 Oct;109(10):2033-2039. doi: 10.1111/apa.15197. Epub 2020 Feb 21.

.

Atac DG, Koller S, Hanson JVM, Feil S, Tiwari A, Bahr A, Magyar I, Kottke R, **Gerth-Kahlert C**, Berger W.
Hum Mol Genet. 2020 Jan 1;29(1):132-148.

Longstanding diplopia after ethmoidal artery ligation for epistaxis.
Bischoff S, **Gerth-Kahlert** C, Holzmann D, Soyka MB.
Eur Arch Otorhinolaryngol. 2020 Jan;277(1):161-167.

2019

Colour vision testing in young children with reduced visual acuity.
Pfäffli OA, Tamási B, Hanson JVM, **Gerth-Kahlert** C.
Acta Ophthalmol. 2020 98(1):e113-e120 [Epub ahead of print2019 Aug 30].

Genotype-Phenotype Analysis of a Novel Recessive and a Recurrent Dominant SNRNP200 Variant Causing Retinitis Pigmentosa.
Gerth-Kahlert C, Koller S, Hanson JVM, Baehr L, Tiwari A, Kivrak-Pfiffner F, Bahr A, Berger W.
Invest Ophthalmol Vis Sci. 2019 1;60(8):2822-2835.

Tuberöse Sklerose Komplex: Analyse des okulären Phänotyps und assoziierte Komplikationen.
Zweipfenning, F, Toelle SP, **Gerth-Kahlert** C.
Klin Monatsbl. Augenheilkunde 2019;236(4):462-468.

2018

Absence of goniodysgenesis in patients with chromosome 13q-microdeletion related microcoria.
Gerth-Kahlert C, Maggi J, Töteberg-Harms M, Tiwari M, Budde B, Nürnberg P, Koller S, Berger W.
Ophthalmology Glaucoma 2018 1(2):145–147.

Prediction of ROP Treatment and Evaluation of Screening Criteria in VLBW Infants-a Population Based Analysis.
Gerull R, Brauer V, Bassler D, Laubscher B, Pfister RE, Nelle M, Müller B, Roth-Kleiner M, **Gerth-Kahlert** C, Adams M; Swiss Neonatal Network & Follow-up Group.
Pediatr Res. 2018 84(5):632-638.

Outcome of Pediatric Cataract Surgeries in a Tertiary Center in Switzerland.
Ambroz S, Töteberg-Harms M, Funk J, Barthelmes D, Hanson JVM, **Gerth-Kahlert** C.
J of Ophthalmol 2018 Feb 25:3230489.

Spontaneous Nystagmus in the Dark in an Infantile Nystagmus Patient May Represent Negative Optokinetic Afternystagmus.
Lin TF, **Gerth-Kahlert** C, Hanson JVM, Straumann D, Huang MY.
Front Neurol. 2018 Mar 14;9:151.

Outer retinal dysfunction in the absence of structural abnormalities in multiple sclerosis.)
Hanson JVM, Hediger M, Manogaran P, Landau K, Hagenbuch N, Schippling S, **Gerth-Kahlert** C.
Invest Ophthalmol Vis Sci 2018 1(59):549-560.

Incidence of ROP and ROP Treatment in Switzerland 2006-2015 – a Population Based Analysis.
Gerull R, Brauer V, Bassler D, Laubscher B, Pfister R, Nelle M, Müller B, **Gerth-Kahlert** C, Adams M.
Arch Dis Child Fetal Neonatal Ed. 2018 103(4):F337-F342.

Retinal ganglion cell topography in patients with visual pathway pathology.
Zehnder S, Wildberger H, Hanson JVM, Lukas S, Pelz S, Landau K, Wichmann W, **Gerth-Kahlert** C.
J Neuroophthalmol. 2018 38(2):172-178.

2017

Long-term follow up in children with anisocoria: cocaine test results and patient outcome.
Fierz FC, **Gerth-Kahlert** C.
J of Ophthalmol 2017 7575040, Epub 2017 Dec 4.

Infantile hemangiomas with conjunctival involvement: an underreported occurrence.
Theiler M, Baselga E, **Gerth-Kahlert** C, Mathes EF, Weibel L, Hohermuth S, Schwieger-Briel A, Frieden IF.
Pediatric Dermatolol. 2017 34(6):681-685.

C2orf71 mutations as a frequent cause of autosomal-recessive retinitis pigmentosa: clinical analysis and presentation of 9 novel mutations.
Gerth-Kahlert C, Tiwari A, Hanson JVM, Batmanabane V, Traboulsi E, Pennesi ME, Al-Qahtani AA, Lam BL, Heckenlively J, Zweifel SA, Vincent A, Fierz F, Barthelmes D, Branham K, Khan N, Bahr A, Baehr L, Magyar I, Koller S, Azzarello-Burri S, Niedrist D, Heon E, Berger W.
Invest Ophthalmol Vis Sci. 2017 58(10):3840-3850.

Senescent Changes and Topography of the Dark-Adapted Multifocal Electroretinogram.
Panorgias A, Tillman M, Sutter EE, Moshiri A, **Gerth-Kahlert** C, Werner JS.
Invest Ophthalmol Vis Sci. 2017 58(2):1323-1329.

Characterization of two novel intronic OPA1 mutations resulting in aberrant pre-mRNA splicing.
Bolognini R, **Gerth-Kahlert** C, Abegg M, Bartholdi D, Mathis N, Sturm V, Gallati S, Schaller A.
BMC Med Genet. 2017 28;18(1):22.

Clinical Utility Gene Card for Non-syndromic Microphthalmia Including Next Generation Sequencing Based Approaches.
Richardson R, Sowden J, **Gerth-Kahlert** C, Moore AT, Moosajee M.
Eur J Hum Genet. 2017 Apr;25(4). doi: 10.1038/ejhg.2016.201.

2016

Next generation sequencing based identification of disease-associated mutations in Swiss patients with retinal dystrophies.
Tiwari A, Bahr A, Baehr L, Fleischhauer J, Zinkernagel M, Winkler N, Barthelmes D, Berger L, **Gerth-Kahlert** C, Neidhardt J, Berger W.
Scientific Reports 2016 6:28755.

A secreted WNT-ligand-binding domain of FZD5 generated by a frameshift mutation causes autosomal dominant coloboma.
Liu C, Widen SA, Williamson KA, Ratnapriya R, **Gerth-Kahlert** C, Rainger J, Alur RP, Strachan E, Manjunath SH, Balakrishnan A, Floyd JA; UK10K Consortium, Li T, Waskiewicz A, Brooks BP, Lehmann OJ, FitzPatrick DR, Swaroop A.
Hum Mol Genet. 2016 25(7):1382-91.

Biallelic mutations in *CRB1* underlie Autosomal Recessive Familial Foveal Retinoschisis
Vincent A, Ng J, **Gerth-Kahlert** C, Tavares E, Maynes JT, Wright T, Tiwari A, Tumber A, Li S, Hanson JV, Bahr A, MacDonald H, Bähr L, Westall C, Berger W, Cremers FP, den Hollander AI, Héon E.
Invest Ophthalmol Vis Sci 2016 57:2637–2646.

2015

Intra-familial phenotype variability in patients with Jalili syndrome.
Gerth-Kahlert C, Seebauer B, Dold S, Hanson JV, Wildberger H, Spörri A, van Waes H, Berger W.
Eye. 2015 29(5):712-6.

Boucher-Neuhäuser syndrome: cerebellar degeneration, chorioretinal dystrophy and hypogonadotropic hypogonadism: two novel cases and a review of 40 cases from the literature.
Tarnutzer AA, **Gerth-Kahlert** C, Timmann D, Chang DI, Harmuth F, Bauer P, Straumann D, Synofzik M.
J Neurol. 2015 262 (1): 194-202.

2013

Dental Phenotype in Jalili Syndrome due to a c.1312 dupC Homozygous Mutation in the CNNM4 Gene.
Luder HU, **Gerth-Kahlert** C, Ostertag-Benzinger S, Schorderet DF.

PLOS ONE, 2013 Oct 23, 8(10): e78529.

Clinical and mutation analysis of 51 probands with anophthalmia and/ or severe microphthalmia from a single center.
Gerth-Kahlert C, Williamson K, Ansari M, Rainger JK, Hingst V, Zimmermann T, Tech S, Guthoff RF, van Heyningen V, FitzPatrick DR.
Mol Genet & Genomic Med, 2013. 1(1): 15-31

2011

BBS genotype-phenotype assessment of a multiethnic patient cohort calls for a revision of the disease definition.
Deveault C, Billingsley G, Duncan JL, Bin J, Theal R, Vincent A, Fieggen KJ, **Gerth** C, Noordeh N, Traboulsi EI, Fishman GA, Chitayat D, Knueppel T, Millán JM, Munier FL, Kennedy D, Jacobson SG, Innes AM, Mitchell GA, Boycott K, Héon E.
Hum Mutat. 2011 32: 610-619.

Mutations in chaperonin-like BBS genes are a major contributor to disease development in a multiethnic Bardet-Biedl syndrome patient population.
Billingsley G, Bin J, Fieggen KJ, Duncan JL**, Gerth C**, Ogata K, Wodak SS, Traboulsi EI, Fishman GA, Paterson A, Chitayat D, Knueppel T, Millán JM, Mitchell GA, Deveault C, Héon E.
J Med Genet. 2011. 47(7):453-63.

2009

High-resolution retinal imaging of young children using a hand-held scanner and Fourier-domain OCT.
Gerth C, Zawadzki RJ, Werner JSW, Heon E.
JAAPOS 2009. 13:72-74.

Retinal Microstructure in patients with EFEMP1 retinal dystrophy evaluated by Fourier-domain OCT.
Gerth C, Zawadzki RJ, Werner JSW, Heon E.
Eye 2009. 23:480-483.

2008

Ocular phenotypic in patients with methylmalonic aciduria and homocystinuria, cobalamin C type.
Gerth *C,* Morel CF, Feigenbaum A, Levin AV.
JAAPOS 2008 12:591-596.

Timing of Surgery for Infantile Esotropia in Humans: Effects on Cortical Motion Visual Evoked Responses.
Gerth C, Mirabella G, Li X, Wright T, Westall CA, Colpa L, Wong A
Invest Ophthalmol Vis Sci. 2008. 49:3432-3437.

A comparison of signal detection techniques in the multifocal electroretinogram.
Wright T, Nielssen J, **Gerth** *C,* Westall C.
Doc Ophthalmol. 2008.117:163-170.

Retinal morphology of patients with X-linked Retinoschisis evaluated by Fourier-Domain Optical Coherence Tomography.
Gerth C, Zawadzki RJ, Werner JSW, Heon E.
Arch Ophthalmol. 2008.126:807-811.

Gerth C, Zawadzki RJ, Werner JSW, Heon E.
Retinal morphology in patients with BBS1 and BBS10 related Bardet–Biedl Syndrome evaluated by Fourier-domain optical coherence tomography. (2008)
Vis Res. 48: 392-399

2007

Novel RDH12 Mutations Associated with Leber Congenital Amaurosis and Cone-Rod Dystrophy: Biochemical and Clinical Evaluations.
Sun W, **Gerth** *C*, Maeda AM, Lodowski, DT, Van Der Kraak L, Saperstein DA, Heon E, Palczewski K.
Vis Res. 2007. 47: 2055–2066.

Gerth C, Wright T, Heon E, Westall CA.
Assessment of central retinal function in patients with advanced retinitis pigmentosa. (2007)
Invest Ophthalmol Vis Sci. 48: 1312-1318.

2006

Clinical Application of Rapid Serial Fourier Domain Optical Coherence Tomography For Macular Alam S, Zawadzki RJ, Choi S, **Gerth** *C*, Park SS, Morse LS, Werner JS.
Imaging.
Ophthalmology 2006. 113: 1425-1431.

Gerth *C*, Delahunt PB, Alam S, Morse LS, Werner JS.
Cone-mediated MfERG in AMD: progression over a long-term follow-up. (2006)
Arch Ophthalmol. 124:345-352.

Photodynamic therapy for choroidal neovascularization in patients with multifocal choroiditis and panuveitis.
Gerth *C,* Spital G, Lommatzsch A, Heiligenhaus A, Pauleikhoff D.
Eur J Ophthalmol 2006. 16(1):111-8.

2004

Senescence of human multifocal electroretinogram (mfERG) components - a localized approach.
Tzekov R, **Gerth** *C*, Werner JS.
Graefe's Clin Arch Exp Ophthalmol. 2004. 7:549-560.

Lutein, Zeaxanthin, Macular Pigment and Visual Function in Adult Cystic Fibrosis Patients.
Schupp CI, Olano-Martin E, **Gerth** C, Morrissey BM, Cross CE, Werner JS.
Am J Clin Nutr. 2004. 79:1045-1052.

2003

Gerth *C*, Hauser D, Delahunt PB, Morse LS, Werner JS.
Assessment of multifocal electroretinogram abnormalities and their relation to morphologic characteristics in patients with large drusen.
Arch Ophthalmol. 2003. 121: 1404-1414.

MfERG response dynamics of the aging retina.
Gerth *C*, Sutter EE, Werner JS.
Invest Ophthalmol Vis Sci. 2003. 44: 4443-4450.

Topography of the chromatic pattern-onset VEP.
Gerth *C*, Delahunt PB, Crognale MA, Werner JS.
Journal of Vision 2003. 3: 171-182

2002

Gerth *C*, Garcia SM, Ma L, Keltner JL, Werner JS.
Multifocal ERG: Age-related changes for different luminance levels.
Graefe's Clin Arch Exp Ophthalmol. 2002. 240: 202-208.

Phenotypes of 16 Stargardt/FFM disease patients with known ABCA4 mutations and evaluations of genotype-phenotype correlation.
Gerth *C*, Andrassi-Darida M, Bock M, Preising M, Weber B, Lorenz B.
Graefe's Clin Arch Exp Ophthalmol. 2002. 240: 628-638.

2000

Early- onset severe rod-cone dystrophy in young children with RPE65 mutations.
Lorenz B, Gyürüs P, Preising M, Bremser D, Gu S, Andrassi M, **Gerth** *C*, Gal A.
Invest Ophthalmol Vis Sci. 2000. 41: 2735-2742.

Impact of notch filter use on waveforms of first- and second-order-kernel responses from multifocal ERGs.
Bock M, **Gerth** *C*, Lorenz B.
Doc Ophthalmol. 2000. 101:195-210.

Case reports/ commentary

2020

[A different lens reflex, what to do next?]
Pfäffli OA, **Gerth-Kahlert** C.
Ophthalmologe. 2020 Mar 3. doi: 10.1007/s00347-020-01055-z. [Epub ahead of print].

Outer Retinal Dysfunction on Multifocal Electroretinography May Help Differentiating Multiple Sclerosis From Neuromyelitis Optica Spectrum Disorder.
Hanson JVM, Schippling S, **Gerth**-Kahlert C
Front Neurol. Apr 2020. 28;11:282.

2018

Ungewöhnlicher kongenitaler Irisbefund.
Seiler R, Gunzinger JM, Rüschoff JH, Barthelmes D, Bode PK, **Gerth-Kahlert** C.
Ophthalmologe, 2018 115 (3):235-238.

2017

Unusual retinopathy in a child with severe combined immune deficiency.
Gerth-Kahlert C, Tiwari A, Hauri-Hohl M, Hanson JVM, Bahr A, Palmowski-Wolfe A, Güngör T, Berger W.
Ophthalmic Genet. 2017 Aug 16:1-3.

Horizontal Gaze Palsy in Two Brothers with Compound Heterozygous ROBO3 Gene Mutations.
Hackenberg A, Boltshauser E, **Gerth-Kahlert** C, Stahr N, Azzarello-Burri S, Plecko B.
Neuropediatrics. 2017 48(1):57-58.

2016

Teaching NeuroImages: Recurrent oculomotor palsies caused by neurosarcoidosis.
Kana V, Petersen JA, Ikenberg K, Chappaz A, **Gerth-Kahlert** C, Appenzeller P, Linnebank M.
Neurology 2016 87(3): e31-2.

Iris cyst in a child with Aicardi syndrome: a novel association.
Chappaz A, Barthelmes D, Buser L, Funk J, **Gerth-Kahlert** C.
J AAPOS. 2016 20(5):451-452.

Maculopathy following exposure to visible and infrared radiation from a laser pointer: a clinical case study.
Hanson JV, Sromicki J, Mangold M, Golling M, **Gerth-Kahlert** C.
Doc Ophthalmol. 2016 132(2):147-55.

2008

Gerth C, Zawadzki RJ, Licht C, Werner JSW, Heon E.
A Microstructural retinal analysis of membrano-proliferative glomerulonephritis type II.
BJO 2008. 92: 1150-1160.

Reviews

2018

Ziliopathien.
Gerth-Kahlert C, Koller S.
Klin Monbl Augenheilkd. 2018 Mar;235(3):264-27.

2016

Optical coherence tomography and magnetic resonance imaging in multiple sclerosis and neuromyelitis optica spectrum disorder.
Manogaran P, Hanson JVM, Olbert E, Egger C, Wicki C, **Gerth-Kahlert** C, Landau K, Schippling S.
Int J Mol Sci. 2016 17(11). pii: E1894.