Study Number	Paper Title	First Author	Year of Publication	Journal	PMID	Country of Study	Total Study Cohort*	Included in this Study	Condition
1	Evidence of macular pigment in the central macula in albinism	Wolfson Y et al.,	2016	Experimental Eye Research	26474496	USA	4	3	Albinism
2	The ophthalmic presentation of Hermansky-Pudlak syndrome 6	Hull S et al.,	2016	British Journal of Ophthalmology	26823395	India	3	3	Albinism
3	A Japanese Family With Autosomal Dominant Oculocutaneous Albinism Type 4	Oki R et al.,	2016	Investigative Ophthalmology and Visual Science	28192564	Japan	16	14	Albinism
4	Molecular genetic and clinical evaluation of three Chinese families with X-linked ocular albinism	Zou X et al.,	2017	Scientific Reports	28211458	China	4	3	Albinism
5	The Henle Fiber Layer in Albinism: Comparison to Normal and Relationship to Outer Nuclear Layer Thickness and Foveal Cone Density	Lee DJ et al.,	2018	Investigative Ophthalmology and Visual Science	30398625	USA	38	4	Albinism
6	Bilateral atypical lamellar holes in a patient with oculocutaneous albinism	Falcone MM et al.,	2020	Ophthalmic Genetics	32543925	UK	1	1	Albinism
7	PAX6 Genotypic and Retinal Phenotypic Characterization in Congenital Aniridia	Pedersen HR et al.,	2020	Investigative Ophthalmology and Visual Science	32396632	Norway	95	23	PAX6
8	Correlation of novel PAX6 gene abnormalities in aniridia and clinical presentation	Sannan NS et al.,	2017	Canadian Journal of Ophthalmology	29217025	Canada	33	25	PAX6
9	PAX6 missense variants in two families with isolated foveal hypoplasia and nystagmus: evidence of paternal postzygotic mosaicism.	Cunha DL et al.,	2021	European Journal of Human Genetics	33024313	UK	6	4	PAX6
10	Autosomal dominant foveal hypoplasia without visible macular abnormalities and PAX6 mutations.	Matsushita I et al.,	2020	Japanese Journal of Ophthalmology	32857266	Japan	4	4	PAX6
11	Nonsense suppression induced readthrough of a novel PAX6 mutation in patient-derived cells of congenital aniridia.	Liu X et al.,	2020	Molecular Genetics and Genomic Medicine	32125788	China	12	1	PAX6
12	Impaired DNA-binding affinity of novel PAX6 mutations.	Lee S et al.,	2020	Scientific Reports	32080308	South Korea	5	5	PAX6
13	Recurrent PAX6 mutation in a Chinese family with congenital aniridia, progressive cataracts and mental retardation.	Chen DD et al.,	2020	European Journal of Ophthalmology	30426773	China	11	4	PAX6
14	Aniridia due to a novel microdeletion affecting PAX6 regulatory enhancers: case report and review of the literature.	Syrimis A et al.,	2018	Journal of Genetics	29932076	Cyprus	2	2	PAX6
15	Novel PAX6 mutation reported in an aniridia patient.	Winegarner A et al.,	2017	Human Genome Variation	29238604	Japan	1	1	PAX6
16	Newly identified paired box 6 mutation of variant familial aniridia: Congenital iris ectropion with foveal hypoplasia.	Kim WJ et al.,	2017	Indian Journal of Ophthalmology	28300742	South Korea	2	2	PAX6
17	A novel de novo duplication mutation of PAX6 in a Chinese family with aniridia and other ocular abnormalities	Zhuang J et al.,	2014	Scientific Reports	24787241	China	5	1	PAX6
18	The Role of Optical Coherence Tomography in an Atypical Case of Oculocutaneous Albinism: A Case Report	Rossi S et al.,	2012	Case Reports in Ophthalmology	22548044	Italy	1	1	Albinism
19	A novel splicing site mutation of the GPR143 gene in a Chinese X-linked ocular albinism pedigree	Cai CY et al.,	2013	Genetics and Molecular Research	24301936	China	1	1	Albinism
20	A new phenotype of recessively inherited foveal hypoplasia and anterior segment dysgenesis maps to a locus on chromosome 16q23.2-24.2	Pal B et al.,	2014	Journal of Medical Genetics	15466012	UK	16	1	SLC38A8
21	A new recessively inherited disorder composed of foveal hypoplasia, optic nerve decussation defects and anterior segment dysgenesis maps to chromosome 16q23.3-24.1	Al-Araimi M et al.,	2013	Molecular Vision	24194637	Pakistan	7	3	SLC38A8
22	Novel Biallelic Variants and Phenotypic Features in Patients with SLC38A8-Related Foveal Hypoplasia	Schiff ER et al.,	2021	International Journal of Molecular Sciences	33498813	UK	9	6	SLC38A8
23	Structural modeling of a novel SLC38A8 mutation that causes foveal hypoplasia	Toral MA et al.,	2017	Molecular Genetics and Genomic Medicine	28546991	USA	2	2	SLC38A8
24	Genetic causes of nystagmus, foveal hypoplasia and subnormal visual acuity- other than albinism	Ehrenberg M et al.,	2021	Ophthalmic Genetics	33594928	Israel	31	4	SLC38A8
25	The pathogenicity of SLC38A8 in five families with foveal hypoplasia and congenital nystagmus	Weiner C et al.,	2020	Experimental Eye Research	32032626	Israel	8	1	SLC38A8
26	Homozygous stop mutation in AHR causes autosomal recessive foveal hypoplasia and infantile nystagmus	Mayer AK et al.,	2019	Brain	31009037	UK	2	2	AHR
27	Correlation between electroretinography, foveal anatomy and visual acuity in albinism	Hu Z et al.,	2019	Advances in Ophthalmology	30927186	USA	35	16	Albinism
28	Clinical and Molecular Characterization of Achromatopsia Patients: A Longitudinal Study	Brunetti-Pierri R et al.,	2021	International Journal of Molecular Sciences	33562422	Italy	21	3	Achromatopsia
29	Long-Term Investigation of Retinal Function in Patients with Achromatopsia	Georgiou M et al.,	2020	Investigative Ophthalmology and Visual Science	32960951	UK	18	17	Achromatopsia
30	Phenotype Driven Analysis of Whole Genome Sequencing Identifies Deep Intronic Variants that Cause Retinal Dystrophies by Aberrant Exonization	Di Scipio M et al.,	2020	Investigative Ophthalmology and Visual Science	32881472	Canada	15	1	Achromatopsia
31	Retinal Structure and Function in Achromatopsia: Implications for Gene Therapy	Sundaram V et al.,	2014	Ophthalmology	24148654	UK	40	36	Achromatopsia
32	Photoreceptor structure and function in patients with congenital achromatopsia	Genead MA et al.,	2011	Investigative Ophthalmology and Visual Science	21778272	USA	12	11	Achromatopsia
33	Diagnostic fundus autofluorescence patterns in achromatopsia	Fahim AT et al.,	2013	American Journal of Ophthalmology	23972307	USA & Germany	10	8	Achromatopsia
34	Spectral-domain optical coherence tomography staging and autofluorescence imaging in achromatopsia	Greenberg JP et al.,	2014	JAMA Ophthalmology	24504161	USA	17	6	Achromatopsia
35	Retinal morphology of patients with achromatopsia during early childhood: implications for gene therapy	Yang P et al.,	2014	JAMA Ophthalmology	24676353	USA	18	8	Achromatopsia
36	Genotype-Dependent Variability in Residual Cone Structure in Achromatopsia: Toward Developing Metrics for Assessing Cone Health	Dubis AM et al.,	2014	Investigative Ophthalmology and Visual Science	25277229	UK	18	4	Achromatopsia

37	Congenital Achromatopsia and Macular Atrophy Caused by a Novel Recessive PDE6C Mutation (p.E591K)	Katagiri S et al.,	2015	Ophthalmic Genetics	25605338	Japan	4	2	Achromatopsia
38	Novel CNGA3 mutations in Chinese patients with achromatopsia	Lang X et al.,	2015	British Journal of Ophthalmology	25637600	China	15	13	Achromatopsia
39	Novel mutations in the gene for α-subunit of retinal cone cyclic nucleotide-gated channels in a Japanese patient with congenital achromatopsia	Kuniyoshi K et al.,	2016	Japanese Journal of Ophthalmology	27040408	UK	1	1	Achromatopsia
40	Residual Foveal Cone Structure in CNGB3-Associated Achromatopsia	Langlo CS et al.,	2016	Investigative Ophthalmology and Visual Science	27479814	USA	51	5	Achromatopsia
41	In vivo imaging of a cone mosaic in a patient with achromatopsia associated with a GNAT2 variant	Ueno S et al.,	2017	Japanese Journal of Ophthalmology	27718025	Japan	1	1	Achromatopsia
42	Repeatability and longitudinal assessment of foveal cone structure in CNGB3-associated achromatopsia	Langlo CS et al.,	2017	Retina	28145975	USA	41	3	Achromatopsia
43	The clinical phenotype of CNGA3-related achromatopsia: pretreatment characterisation in preparation of a gene replacement therapy trial	Zobor D et al.,	2017	Investigative Ophthalmology and Visual Science	28159970	Germany	36	34	Achromatopsia
44	Multimodal imaging including semiquantitative short-wavelength and near-infrared autofluorescence in achromatopsia	Metet A et al.,	2018	Scientific Reports	29618791	France and Germany	16	16	Achromatopsia
45	Novel causative variants in patients with achromatopsia	Abdelkader E et al.,	2018	Ophthalmic Genetics	30289319	Egypt	7	3	Achromatopsia
46	Longitudinal Assessment of Retinal Structure in Achromatopsia Patients With Long-Term Follow-up	Hirji N et al.,	2018	Investigative Ophthalmology and Visual Science	30513534	UK	50	21	Achromatopsia
47	Adaptive Optics Retinal Imaging in CNGA3-Associated Achromatopsia: Retinal Characterization, Interocular Symmetry, and Intrafamilial Variability	Georgiou M et al.,	2019	Investigative Ophthalmology and Visual Science	30682209	UK	38	18	Achromatopsia
48	Characterization of Retinal Structure in ATF6-Associated Achromatopsia	Mastey RR et al.,	2019	Investigative Ophthalmology and Visual Science	31237654	USA	7	7	Achromatopsia
49	Deep Phenotyping of PDE6C-Associated Achromatopsia	Georgiou M et al.,	2019	Investigative Ophthalmology and Visual Science	31826238	UK	8	7	Achromatopsia
50	Macular maldevelopment in ATF6-mediated retinal dysfunction	Ritter M et al.,	2019	Ophthalmic Genetics	31900015	UK	2	2	Achromatopsia
51	Photoreceptor Structure in GNAT2-Associated Achromatopsia	Georgiou M et al.,	2020	Investigative Ophthalmology and Visual Science	32203983	UK	9	4	Achromatopsia
52	Multiexon deletion alleles of ATF6 linked to achromatopsia	Lee E-J et al.,	2020	JCI Insight	32271167	USA	3	1	Achromatopsia
53	Mutations in the unfolded protein response regulator ATF6 cause the cone dysfunction disorder achromatopsia	Kohl S et al.,	2015	Nature Genetics	26029869	Germany	18	15	Achromatopsia

*Total study cohort includes all subjects (disease and control groups) in the original study irrespective of whether there is OCT imaging data/molecular diagnosis.