

Microcornea-Cataract syndrome

Author: Doctors Alrun Gronemeyer¹, Berthold Seitz, Gottfried Naumann

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¹Department of Ophthalmology, University of Erlangen-Nürnberg, Schwabachanlage 6, 91054 Erlangen, Germany. Alrun.Gronemeyer@augen.imed.uni-erlangen.de

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Abstract

Microcornea-cataract syndrome is an autosomal dominant inherited disease characterized by the association of congenital cataract and microcornea without any other systemic anomaly or dysmorphism. Clinical findings include a corneal diameter inferior to 10 mm in both meridians in an otherwise normal eye, and an inherited cataract which is most often bilateral posterior polar with opacification in the lens periphery. The cataract progresses to form a total cataract after visual maturity has been achieved, requiring cataract extraction in the first to third decade of life. Microcornea-cataract syndrome can be associated with other rare ocular manifestations, including myopia, iris coloboma, sclerocornea and Peters anomaly. Ophthalmological and general examination exclude other syndromes. To date, only six families showing microcornea-cataract syndrome have been described. Transmission seems to be autosomal dominant, sometimes with a high degree of penetrance. The gene and the gene locus for this syndrome have not yet been identified. Cataract surgery has to be performed in order to restore visual acuity and avoid amblyopia. Visual acuity after uncomplicated cataract extraction is relatively good.

Keywords

microcornea, cataract, autosomal dominant

Disease name and synonyms

Microcornea-cataract syndrome or Cataract-microcornea syndrome

Excluded diseases

Isolated cataracts, cataract-dental syndrome (Nance-Horan syndrome), Cornea plana, x-linked congenital cataract with microcornea or slight microphthalmia, [microphthalmia](#), [Peters anomaly](#), [Rothmund-Thompson syndrome](#), [Warburgs Micro syndrome](#), [Schwartz-Jampel](#)

[syndrome](#), [WAGR](#) (Wilms tumor, aniridia, genitourinary anomalies, mental retardation) syndrome, [Nail-Patella syndrome](#), oculo-dento-digital dysplasia [dominant](#) or [recessive](#)(1), [acro-reno-ocular syndrome](#), [fronto-facio-nasal dysostosis](#).

Diagnostic criteria /definition

Microcornea-cataract syndrome is an anterior eye dystrophy characterized by the association of congenital cataract and microcornea without any other systemic anomaly or dysmorphism.

Other rare ocular manifestations include myopia, iris coloboma, sclerocornea and Peters anomaly. Transmission seems to be autosomal dominant, sometimes with a high degree of penetrance.

Differential diagnosis

It may be difficult to differentiate clinically the microcornea-cataract syndrome from congenital cataract with posterior sutural opacities in heterozygotes and the cataract-dental syndrome (Nance-Horan). If a pedigree is available, these two syndromes can be differentiated from microcornea-cataract syndrome on the basis of their X-linked transmission.

Frequency

Microcornea-cataract syndrome is a rare disease. Six families showing this syndrome have been described so far (2-7). Taking into account that differential diagnosis of the above mentioned syndromes is not always straightforward, a total of less than 20 families have been reported in the literature so far (10).

Clinical description

Microcornea is characterized by a corneal diameter of less than 10 mm in both meridians in an otherwise normal eye. Differentiation has to be made between microcornea and microphthalmos, in which the whole eye is small. The inherited cataract is most often a bilateral posterior polar cataract with opacification in the lens periphery. However, posterior subcapsular and anterior polar types have been described (6). The cataract progresses to form a total cataract after visual maturity has been achieved, requiring cataract extraction in the first to third decade of life. Visual acuity after uncomplicated cataract extraction is relatively good.

Mollica et al. have described a family with microcornea-cataract syndrome associated with myopia (4).

A 7-generation family reported by Salmon presented with microcornea and cataract in 18 members of the family, an additional 6 members of the family had manifestations of anterior segment mesenchymal dysgenesis. Peripheral sclerocornea with cornea plana, total sclerocornea and Peters' anomaly was seen. This was interpreted as a demonstration of variable expressivity of the gene (6). Green and Johnson also described a family with individuals having Peters anomaly and sclerocornea (3). These patients often presented with other ocular abnormalities such as esotropia, nystagmus, microphthalmus, glaucoma and posterior embryotoxon. Such an affection may decrease the visual prognosis drastically.

Neither dental abnormalities or dysmorphic features nor mental retardation have been described with this syndrome.

Management including treatment

A detailed family history reflecting an autosomal dominant transmission helps define the syndrome. Furthermore, a complete medical (including dental) examination should exclude other syndromes associated with microcornea and cataract.

To restore visual acuity, cataract extraction is necessary sooner or later in all cases. In general, the visual outcome after surgery is good. Some authors point out that the risk of secondary glaucoma following pars plana lensectomy for congenital cataract seems to be high, especially when microcornea is present (9, 11). Therefore careful follow-up is needed. Seitz and Naumann reported successful cataract extraction via ipsilateral autologous keratoplasty in cases of severe microcornea (8).

Etiology

Microcornea-cataract syndrome -a dysplastic malformation of the anterior segment of the eye- is transmitted as an autosomal dominant trait in contrast to X-linked congenital cataract with microcornea or slight microphthalmia. Stefaniak et al. reported a family in which the proportion of affected family members was so high that he suspected a preferential transmission of the chromosome carrying the mutant gene (7). To date, the causative gene for microcornea-cataract syndrome and the gene locus have not been identified.

Salmon suggested that the embryological origins of microcornea, sclerocornea and Peters anomaly are similar on the basis of the analysis of a family with microcornea-cataract syndrome in which 6 individuals had sclerocornea and Peters anomaly (6).

Biological diagnostic tests

No diagnostic tests are currently available.

Genetic counseling

The offspring of an affected patient is at least at 50% risk of being affected with microcornea-cataract syndrome because of the autosomal dominant transmission pattern of the disorder and the preferential transmission of the chromosome carrying the mutant gene, which has been reported in some families (7).

Unresolved questions

Although cataract surgery, even in small children, has become more and more successful, due to the improvement in

microsurgical techniques, the problem of aphakia and amblyopia and how to correct it remains a major challenge in treating congenital cataracts.

Further genetic studies are necessary to identify the causative gene for microcornea-cataract syndrome.

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