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Macular corneal dystrophy – a case report

Macular corneal dystrophy (MCD) also known as Groenouw II dystrophy is an autosomal recessive disease characterized by abnormal deposition of glycosaminoglycans in corneal stroma (3, 7). This type of stromal corneal dystrophy is the most rare in patients. MCD can be classified into three immunophenotypes: type I, IA and II. In type I and IA one does not observe the keratan sulphate antigen (aKS) in serum and in deposits in the cornea, whereas in type II it is present (4). Hereditary pattern is autosomal recessive. The causative gene for MCD CHST6 is localized on the chromosome 16q22. It codes the corneal enzyme N-acetyloglucosamine-6-O-sulphotransferase (GlcNAc6ST). This enzyme catalyzes the sulphation of keratan sulphate, an important component of corneal proteoglycans (1, 6, 8, 10, 11, 12). Macular dystrophy is characterized by multiple, gray-white opacities that are present in the corneal stroma and extend into the peripheral cornea. These stromal opacities are distributed throughout the cornea without clear spaces. MCD involves the entire thickness of cornea and is more superficial centrally and deeper peripherally. The central cornea in this condition may be thinned. Significant cornea guttata may be present (3, 7). The disease begins in the first decade of life, with no sexual predilection. Patients usually have blurred vision which worsens with time. Up to the ages of 20 to 40 vision can be significantly reduced. The most common treatment is keratoplasty, but in some cases changes in the cornea may occur again after surgical intervention (4, 9).

CASE REPORT

A 42-year-old man was referred to the First Department of Ophthalmology in Lublin in July 2005 for severe corneal opacities in both eyes which have substantially impaired his vision for at least ten years (Fig. 1). Before that, his sister had underwent penetrating keratoplasty in both eyes due to macular dystrophy. Best corrected visual acuity in his right eye was counting fingers from 4 meters and in his left eye counting fingers from 2 meters. Intraocular pressure measured was 18 and 17 mmHg respectively. In September 2005 our patient underwent penetrating keratoplasty in his left eye. Corneal graft was closed with two diagonal sutures. After surgical intervention he was administered topical Tobramycin three times daily and Prednisolon five times daily to the left eye. After one week his vision improved to 0.15 on the Snellen chart (Fig. 2).

In January 2006 vision acuity in the right eye of our patient worsened to counting fingers from 1 meter and in the left eye the man achieved 1.0 on the Snellen chart with spherical correction –0.75 dioptres and cylinder correction –3.5 dioptres in axis 35°. Topically he received Prednisolon once daily and artificial tears five times daily to his left eye (Fig. 3).

The patient is planned to undergo penetrating keratoplasty in his right eye as well.

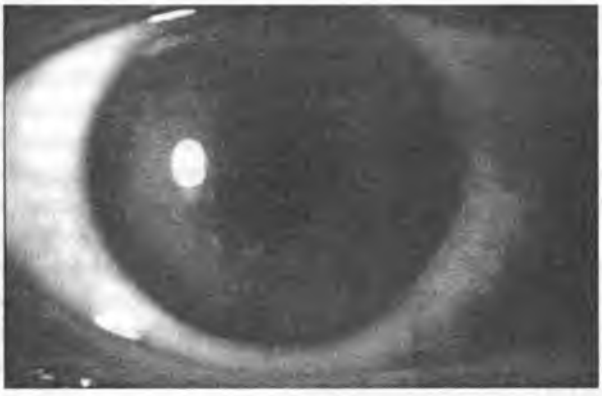

Fig. 1. Left eye of the patient before keratoplasty. BCVA=counting fingers from 2 m

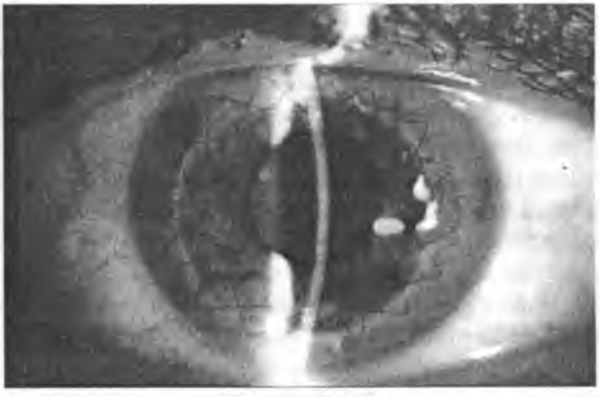

Fig. 2. One week after keratoplasty. BCVA=0.15

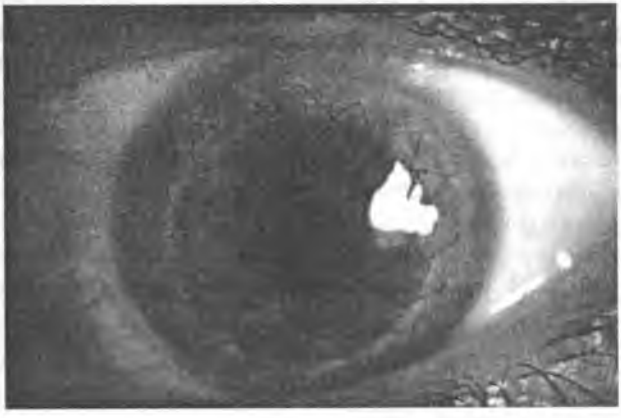

Fig. 3. Four months after keratoplasty. BCVA=1.0

DISCUSSION

Nowadays giving the correct diagnosis of corneal dystrophy consists of many stages. First of all family history should be gained while looking for similar diseases in relatives of the patient. Undergoing clinical examination in a slit lamp is necessary. Genetic examination made from DNA obtained from the peripheral blood such as investigating chromosome 16q22 and looking for gene that codes the corneal enzyme N-acetyloglucosamine-6-O-sulphotransferase become more popular (1, 6, 8, 10, 11, 12). Test ELISA can be useful to determine the level of sulphation of keratan sulphate even if there is no clinical manifestation of the disease (7). In type I MCD there is no keratan sulphate in the cornea, blood and cartilages. In type II there is only 70% of the normal amount of keratan sulphate. If it is possible, for example after penetrating keratopasty, the cornea should undergo histopathologic and immunohistochemical examination. Opacities of the cornea are made of glycosaminoglycans. They are accumulated in endoplasmic reticulum (3).

Up to now there were described only a few methods of treatment of macular corneal dystrophy. If erosions appear they are treated with artificial tears, antibiotics if needed and contact lenses. In patients with low visual acuity authors indicate penetrating keratoplasty as the effective way of improving it (2, 9, 11). Al-Swailem and all presented the results of penetrating keratoplasty in 229 eyes with macular dystrophy (2). Mean follow-up period was 5.9±3.8 years, the mean best corrected visual acuity was 20/50. However among postoperative complications was graft failure, which occurred in 10%. There was a statistically significant increased probability of graft survival if the patient was younger than 40. Besides, corneal endothelial rejections appeared in 20% of eyes, microbial corneal inflammation in 6.1% and recurrence in 5.2%.

Another method is phototherapeutic keratectomy and according to long-term results of this procedure presented by Hafner et al. (5) or a case reported by Wagoner et al. (13) it can increase BCVA moderately for a limited period of time. Defining specific mutations in gene CHST6 can have influence on genetic therapy of this disease in the future.

CONCLUSIONS

Penetrating keratoplasty can be an effective method of treatment of macular dystrophy of the cornea and patients can achieve excellent best corrected visual acuity. However, according to the previous publications graft rejection, microbial corneal inflammation, irregular astigmatism and recurrence of corneal opacities due to macular corneal dystrophy may appear.

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REFERENCES

1. Akama T. O. et al.: Macular corneal dystrophy type I and type II are caused by distinct mutations in a new sulphotransferase gene. Nat. Genet., 26 (2), 237, Oct. 2000.
2. Al-Swailem S. A. et al.: Penetrating keratoplasty for macular corneal dystrophy. Ophthalmology, 112 (2), 220, Feb. 2005.
3. Basic and Clinical Science Course "External Disease and Cornea", Section 8. Polish edition: J. Szaflik, Wyd. Med. Urban & Partner, Wrocław 2004.

4. Cursiefen C. et al.: Immunohistochemical classification of primary and reccurent macular corneal dystrophy in Germany: subclassification of immunophenotype IA using a novel keratan sulfate antibody. Exp. Eye Res., 73 (5), 593, Nov. 2001.
5. Hafner A. et al.: Long-term results of phototherapeutic keratectomy with 1993-nm excimer laser for macular corneal dystrophy. Am. J. Ophthalmol., 140 (3), 392, 2005.
6. Hasegawa N. et al.: Decreased GlcNAc6-O-sulfotransferase activity in the cornea with macular corneal dystrophy. Invest. Ophthalmol. Vis. Sci., 41 (12), 3670, Nov. 2000.
7. Kanski J. J.: Okulistyka kliniczna. Wyd. Med., Wrocław 2005.
8. Klintworth G. K. et al.: Macular corneal dystrophy. Lack of keratan sulfate in serum and cornea. Ophthalmic Paediatr. Genet., 7 (3), 139, Dec. 1986.
9. Klintworth G. K.: Recurrence of macular dystrophy within grafts. Am. J. Ophthalmol., 95 (1), 60, 1983.
10. Liu N. P. et al.: Mutations in corneal carbohydrate sulfotransferase 6 gene (CHST6) cause macular corneal dystrophy in Iceland. Mol. Vis., 13 (6), 261, Dec. 2000.
11. Mrukwa-Kominek E. et al.: Macular corneal dystrophy – clinical state, histopathologic, immunohistochemical examinations and genetical dependence. Klin. Oczna, 101 (6), 802, 2004.
12. Plaas A. H. et al.: Altered fine structures of corneal and skeletal keratan sulfate and chondroitin/dermatan sulfate in macular corneal dystrophy. J. Biol. Chem., 26, 276 (43), 39788, Oct. 2001.
13. Wagoner M. D., Badr I. A.: Phototherapeutic keratectomy for macular corneal dystrophy. J. Refract. Surg., 15 (4), 481, 1999.
14. Wylęgała E. et al.: Macular corneal dystrophy including histologic and ultrastructural changes. Klin. Oczna, 106 (1-2), 68, 2004.

SUMMARY

The aim of this study was to present a case report of macular corneal dystrophy. A case history and other medical documents of a patient referred to the First Eye Hospital in Lublin in July 2005. In July 2005 best corrected visual acuity before surgical intervention in the right eye was counting fingers from 4 m and in the left eye counting fingers from 2 m. Intraocular pressure measured was 18 and 17 mmHg respectively. In September 2005 our patient underwent penetrating keratoplasty in his left eye. Corneal graft was closed with two diagonal sutures. After one week his vision improved to 0.15 on the Snellen chart. In January 2006 vision acuity in the right eye of this patient worsened to counting fingers from 1 m and in the left eye the man achieved 1.0 on the Snellen chart with spherical correction –0.75 dioptres and cylinder correction –3.5 dioptres in axis 35°. Penetrating keratoplasty can be an effective method of treatment of macular dystrophy of the cornea and patients can achieve excellent, best corrected visual acuity. However, according to the previous publications, graft rejection, microbial corneal inflammation, irregular astigmatism and recurrence of corneal opacities due to macular corneal dystrophy may appear.

Dystrofia plamkowa rogówki – opis przypadku

Celem pracy było przedstawienie przypadku dystrofii plamkowej rogówki. Posłużono się historią choroby oraz inną dokumentacja medyczną chorego przyjętego do I Kliniki Okulistyki w Lublinie. W lipcu 2005 r. przed zabiegiem operacyjnym najlepsza ostrość wzroku z korekcją wynosiła liczenie palców z 4 m w prawym oku i liczenie palców z 2 m w oku lewym. Ciśnienie wewnątrzgałkowe

wynosiło odpowiednio 18 i 17 mmHg. We wrześniu 2005 r. u pacjenta wykonano przeszczep drążący rogówki w oku lewym. Na płatek przeszczepu założono szew podwójny diagonalny. Po tygodniu od zabiegu ostrość wzroku poprawiła się do 0.15 na tablicy Snellena. W styczniu 2006 r. nastąpiło pogorszenie ostrości wzroku w oku prawym do liczenia palców z 1 m, natomiast w oku lewym pacjent uzyskał na tablicy Snellena 1.0 z korekcją -0.75 dioptrii sferycznych i -3.5 dioptrii cylindrycznych w osi 35°. Keratoplastyka drążąca może być skuteczną metodą leczenia dystrofii plamkowej rogówki, a pacjent może dzięki temu osiągnąć bardzo dobrą ostrość wzroku. Jednak jak wynika z wcześniejszych publikacji, po operacji mogą wystąpić: odrzucenie przeszczepu, bakteryjne zapalenie rogówki, nieregularny astygmatyzm oraz ponowne pojawienie się zmętnień rogówki związanych z dystrofią plamkową.