Brittle cornea, blue sclera, and red hair syndrome (the brittle cornea syndrome)

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SUMMARY A syndrome of red hair, blue sclera, and brittle cornea with recurrent spontaneous perforations is presented in 2 siblings of a Tunisian Jewish family. The genetic transmission of this disorder is autosomal recessive. This is the second description of this syndrome, which should be called the 'brittle cornea syndrome'. This syndrome has so far been reported only in Tunisian Jewish families.

Brittle cornea with spontaneous perforation is a rare disease. It has been described in association with systemic mesodermal disorders such as osteogenesis imperfecta,¹ Marfan syndrome,² and Ehlers-Danlos syndrome.³

This communication describes a 'brittle cornea' syndrome unrelated to systemic mesodermal disorders. The triad of symptoms includes red hair, brittle megalocornea, and blue sclera. The following 2 cases and the 4 other cases reported previously⁴ represent the above syndrome without any systemic manifestations.

The 'brittle cornea' syndrome seems to be an isolated genetic disorder transmitted as an autosomal recessive trait.

Case reports

Two children out of 5 siblings presented the following disease. The parents were first cousins and originated from a Tunisian Jewish family. Both patients had red hair (Figs. 1, 5), blue sclera, and uniform keratoglobus, and extremely thin corneae (Fig. 3) with several leucomata from previous spontaneous perforations (Figs. 4, 7, 8). On physical examination both children appeared otherwise normal. No systemic manifestations were found, and blood examinations gave normal results. The parents and other members of the family had dark hair and no abnormality of the eyes.

Patient 1 was a red-haired boy, 16 years old (Figs. 1, 2). In the past he had a spontaneous perforation of his left cornea, which had been repaired surgically. On examination the right eye had a

visual acuity of 6/60 with the best myopic correction. The cornea was extremely thin with keratoglobus (Fig. 3). The anterior chamber was deep and the lens was clear. No abnormalities were present in the posterior segment. The intraocular pressure was 16 mmHg.

Examination of the left eye revealed a visual acuity of counting fingers at 2 meters. A large corneal scar was present in the centre of the left cornea with anterior iris adhesions (Fig. 4). The lens remained clear. Fundus examination, so far as could be seen, did not reveal any abnormality.

Patient 2 was a red-haired girl, 8 years old (Figs. 5, 6). She had had recurrent perforations of her eyes, which were surgically repaired. The visual acuity was found to be counting fingers at 1-2 meters due to a central corneal adherent leucoma in each eye (Figs. 7, 8). The lenses and posterior segments were normal so far as could be seen.

Family studies. The parents of the children were first cousins. Both were healthy, without any ocular abnormality. Of their 5 children 3 were normal and two affected. All members of the family except the 2 affected children had dark hair and none had blue sclera, bone fractures, or deafness. The examination of the family could not be extensive because most members live in Tunis. However, only the 2 children described above were red-haired and affected by eye disease. Fig. 9 illustrates the family tree.

Discussion

This brother and sister and 4 other cases previously reported⁴ present a triad of symptoms of red hair, blue sclera, and brittle cornea. Systemic mesodermal

drome, for example, deficiency of lysyl hydroxylase activity in cultured fibroblasts has been reported.³⁻⁹ However the deficiency of this enzyme in similar cases has not been confirmed by others.¹⁰⁻¹¹ Unfortunately such biochemical assays were not performed in our cases. However, they did not present bone fractures or deafness as in osteogenesis imperfecta. Nor did they present skin or ligament hyperelastic changes as in Ehlers-Danlos syndrome, or changes in stature similar to the Marfan syndrome.

In our opinion the 2 siblings suffer from a genetic disorder described only in Tunisian Jews, which can best be called the 'brittle cornea syndrome'. This syndrome has 3 major components: spontaneous perforation of cornea (brittle cornea), blue sclera, and red hair. It should be mentioned that red hair is very uncommon in Tunisian Jews and seems to be a 'marker' of the brittle cornea syndrome.

From the family presented here, and the family reported previously⁴ it is evident that this disorder is transmitted as an autosomal recessive trait. The three traits of brittle cornea, blue sclera, and red hair are transmitted by a single gene. All reported homozygotes presented the complete syndrome.

References

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