Congenital stationary nightblindness in a patient with osteopetrosis

CASE REPORT

The patient was a 36-year-old woman whose parents were first cousins. She and two of her three siblings had had nightblindness and high myopia since childhood, although we had no chance to examine her siblings. A chest x-ray showed increased bone density and diffusely sclerotic bone structures. Computed tomography scan showed diffuse thickening and increased density of the skull bones (Fig 1). Narrowing of the optic canals was not observed radiologically. She was diagnosed as having adult onset osteopetrosis. Her visual acuity was 20/25 with a correction of -8.0 D in both eyes. No abnormality was observed in eye movement or in the pupils. Funduscopically, myopic changes were observed. The margins of the optic discs were clear and the colour was good, although the discs were tilted (Fig 2). The dark adaptation curve showed a normal rod-dark adaptation. The light peak/dark trough ratios on electro-oculogram were 2.2-2.

The electroretinogram (ERG) was recorded using contact lens electrodes (Kyoru-Contact Lens, AE, Kyoto, Japan) as active electrodes. The light source was a stroboscopic light (Nihon-Kohden, SLS3100, Tokyo). After 30 minutes of dark adaptation, scotopic b-wave, photopic flickering, and a single white flash responses were recorded with a blue dim light, 30 Hz flickering light, and a white flash light of 40 J, respectively. Single flash ERGs showed negative-type ERG. Scotopic ERGs were extinguished (Fig 2C).

COMMENT

CSNB has been classified into CSNB with abnormal fundus (Opuch's disease and fundus albipunctatus) and CSNB with normal fundus. In CSNB with normal fundus, the fundus appearance is essentially normal, although some patients show changes associated with high myopia or optic disc changes (tilted, pale, or dysplastic). CSNB with normal fundus, which is inherited as an autosomal recessive, autosomal dominant, or X linked recessive trait, is classified into two types. Type I shows a markedly reduced ERG. Type II shows a negative type ERG; a normal a-wave with a markedly reduced or
Accordingly, this patient was examined under anesthesia and found to have a 'thumb-sized' lump on her left upper eyelid over the course of 1 year. Because of a pathological fear of doctors and hospitals she had delayed her presentation until the size of the lump interfered with her wearing of spectacles.

She attended the clinic with a gauze dressing attached to her spectacle frame with which she had been attempting to conceal the mass's increasing bulk. On examination she was found to have a very large tumour, approximately 6 cm long and 3 cm in diameter, attached by a broad base to the left upper eyelid and anterior aspect of the temporal region. Granulation tissue was evident on the surface with a purulent discharge (Fig 1A). There was no evidence of local or systemic metastasis.

She underwent primary diagnostic excision of the tumour, and since the patient insisted upon local anaesthesia and day-case surgery, the tissue defect was left to heal by secondary intention (Fig 1B).

Figure 2 Fundus appearance (A) right eye, (B) left eye). Myopic changes were observed. Optic discs were tilted; however, the colour of the disc was normal. (C) Electroretinogram (ERG). Single white flash ERG showed a negative-type ERG (that is, a normal a-wave with a markedly reduced b-wave). The oscillatory potentials were absent. Scotopic ERG were extinguished in both eyes. Photopic flicker responses were only slightly reduced compared with those of a normal subject.

An unusual impediment to spectacle wear: pilomatrixoma

EDITOR—Pilomatrixoma or calcifying epithelioma of Malherbe is a relatively uncommon benign skin tumour arising from cells of the hair follicle. Sixty per cent present during the first two decades of life although pilomatrixoma can occur at any age.1 It is more common in females by a ratio of 3:2. Although pilomatrixoma has a predilection for the upper lid and eyebrow,2 each ophthalmologist will see a relatively small number. A case of pilomatrixoma is presented with discussion of its management.

CASE REPORT

A 48-year-old woman developed a 'thumb sized' lump on her left upper eyelid over the course of 1 year. Because of a pathological fear of doctors and hospitals she had delayed her presentation until the size of the lump interfered with her wearing of spectacles.

She attended the clinic with a gauze dressing attached to her spectacle frame with which she had been attempting to conceal the mass's increasing bulk. On examination she was found to have a very large tumour, approximately 6 cm long and 3 cm in diameter, attached by a broad base to the left upper eyelid and anterior aspect of the temporal region. Granulation tissue was evident on the surface with a purulent discharge (Fig 1A). There was no evidence of local or systemic metastasis.

She underwent primary diagnostic excision of the tumour, and since the patient insisted upon local anaesthesia and day-case surgery, the tissue defect was left to heal by secondary intention (Fig 1B).

Figure 1 (A) A large 6 cm by 3 cm tumour mass involving the left upper lid and temporal region. The mass demonstrates granulation tissue with a mild purulent discharge. The patient had disguised this mass by means of a gauze dressing attached to her spectacle frame. (B) Appearance of left upper lid and brow 6 months after primary excision of pilomatrixoma allowing large wound defect to heal by secondary intention.

Fig 2A
Fig 2B

Fig 2C

30 Hz Flicker

R

L

Scotopic

15 µV

100 µV

Bright white flash

20 ms

20 ms

Patient

Normal

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an unusual impediment to spectacle wear: pilomatrixoma

30 Hz Flicker

R L

Scotopic

15 µV

100 µV

Bright white flash

20 ms

20 ms

Patient

Normal

When a 48-year-old woman developed a 'thumb sized' lump on her left upper eyelid over the course of 1 year. Because of a pathological fear of doctors and hospitals she had delayed her presentation until the size of the lump interfered with her wearing of spectacles. She attended the clinic with a gauze dressing attached to her spectacle frame with which she had been attempting to conceal the mass's increasing bulk. On examination she was found to have a very large tumour, approximately 6 cm long and 3 cm in diameter, attached by a broad base to the left upper eyelid and anterior aspect of the temporal region. Granulation tissue was evident on the surface with a purulent discharge (Fig 1A). There was no evidence of local or systemic metastasis.

She underwent primary diagnostic excision of the tumour, and since the patient insisted upon local anaesthesia and day-case surgery, the tissue defect was left to heal by secondary intention (Fig 1B).

Figure 1 (A) A large 6 cm by 3 cm tumour mass involving the left upper lid and temporal region. The mass demonstrates granulation tissue with a mild purulent discharge. The patient had disguised this mass by means of a gauze dressing attached to her spectacle frame. (B) Appearance of left upper lid and brow 6 months after primary excision of pilomatrixoma allowing large wound defect to heal by secondary intention.