**Case report by Agnes Choi B.Optom (Hons), M.Optom, GradCertOcTher**

A 17 year old Caucasian female was referred to CFEH following findings of bilateral altered pigmentation at the central macula, reduced vision and nystagmus by her optometrist. Her two younger brothers also have nystagmus and reduced vision.

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Summary: The findings were consistent with cone dystrophy in each eye.

Management plan: Refer to retinal ophthalmologist for review.
Clinical implications

- **Cone dystrophy presents with reduced visual acuity and colour vision abnormalities and may present with photophobia, hemeralopia (poor vision in bright conditions) and/or nystagmus.**

Disorders of cone function can be considered stationary (cone dysfunction syndromes) or progressive (cone and cone-rod dystrophies) [1, 2]. The stationary cone dysfunction syndromes are congenital while progressive cone dystrophies usually present in childhood or early adult life [1, 2]. The stationary subtypes typically have normal rod function whereas the progressive subtypes develop rod photoreceptor dysfunction later in life [2]. Examples of the stationary subtypes include complete and incomplete achromatopsia, and blue cone monochromatism [1, 3].

Reduced visual acuity and colour vision abnormalities typically present in the first or second decades of life in patients with cone dystrophies [4-6]. Cone and cone-rod dystrophies may appear as more severe compared with the rod or rod-cone dystrophies as high acuity vision and the perception of colour are poor [4]. In cone-rod dystrophies, complete blindness occurs in the later stages as the rod photoreceptors undergo degeneration [4, 6]. Cone dystrophies and cone-rod dystrophies can be inherited in an autosomal dominant (20-30%), autosomal recessive (60-70%) or X-Linked recessive (5%) fashion [7,8].

- **Fundus examination, autofluorescence and OCT imaging results may be variable depending on the molecular diagnosis of the cone dystrophy.**

The macula can have a normal to bull’s eye maculopathy appearance while the optic disc may show varying degrees of temporal pallor [6]. The pattern of autofluorescence varies depending on the underlying molecular diagnosis (what mutation and in which gene) and may range from a speckled appearance to focal increased autofluorescence [7]. Abnormalities on OCT imaging typically involve disturbances and attenuation of the EZ (inner segment ellipsoid zone) in the central macula.

- **Reduced or absent light-adapted flash and 30 Hz ERG is characteristic of cone dystrophies.**

Cone function is tested using a single flash (Figure 4) or 30 Hz flicker (Figure 5) stimulation under light-adapted conditions [9]. The light-adapted single flash ERG reflects the cone systems response with information about cone photoreceptor function while the 30 Hz flicker ERG reflects the cone system response as a whole [10]. In the light-adapted single flash ERG waveform, the a-wave arises from cone photoreceptors and cone OFF-bipolar cells while the b-wave comes from ON- and OFF-cone bipolar cells [10]. In cone dystrophies, full-field ERG shows reduced cone responses that worsen with time (in this case both are essentially absent) while rod responses are initially normal (as in Figure 6) [6]. The characteristic ERG responses of relatively preserved dark-adapted response with reduced or absent light-adapted flash and 30 Hz responses is the gold standard for diagnosis [3].

References