An intriguing case of blurred vision in a young patient

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DESCRIPTION

A 25-year-old man with homocystinuria (HCU) presented with a 6-month history of blurred vision. His acuity measured 6/6 (20/20) bilaterally with spectacle correction. Examination found bilateral, inferior subluxation of the crystalline lens with zonular disruption ([figure 1](#)).

DISCUSSION

HCU is an autosomal recessive disorder of methionine metabolism, with an incidence of 1/100 000, resulting in systemic accumulation of homocysteine.

Features include blonde hair, blue irides, marfanoid habitus and arachnodactyly. Neurodevelopmental delay can accompany these features. If suspected, urine screening for cyanide nitroprusside is undertaken. The Guthrie heel-prick test during neonatal screening also detects high methionine levels.

Displacement of the crystalline lens (ectopia lentis), typically occurs inferonasally in untreated HCU by age 25. The lens zonules (suspensory ligaments) contain cysteine but if replaced by homocysteine, disintegration occurs ([figure 2](#)). Retinal detachment and optic atrophy is associated.¹ Raised homocysteine levels can cause carotid stenosis, low vitamin B₁₂ levels and thrombogenesis, making secondary stroke prevention crucial.²

Treatment involves vitamin B₆ supplementation. Ocular management is conservative if symptoms are tolerated. Surgical intervention involves removal of the crystalline lens with intraocular lens implantation. Iris or scleral fixated lenses may be

Figure 2 Magnified image of lens zonules (suspensory ligaments).

considered. Intraocular pressure needs to be controlled if the iridocorneal angle becomes involved.³

Learning points

Systemic pathology must be considered if lens dislocation occurs. As with this case, Snellen visual acuity may not be severely affected. Light entering the peripheral lens instead of the true optical centre causes symptomatic image blurring.

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Competing interests None.

Patient consent Obtained.

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Figure 1 Bilateral ectopia lentis. The zonular suspensory fibres are deficient in cysteine, making them weak and leading to lenticular dislocation.

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