

Images in Medicine

Ectopia lentis and blue sclera in hyperhomocysteinaemia

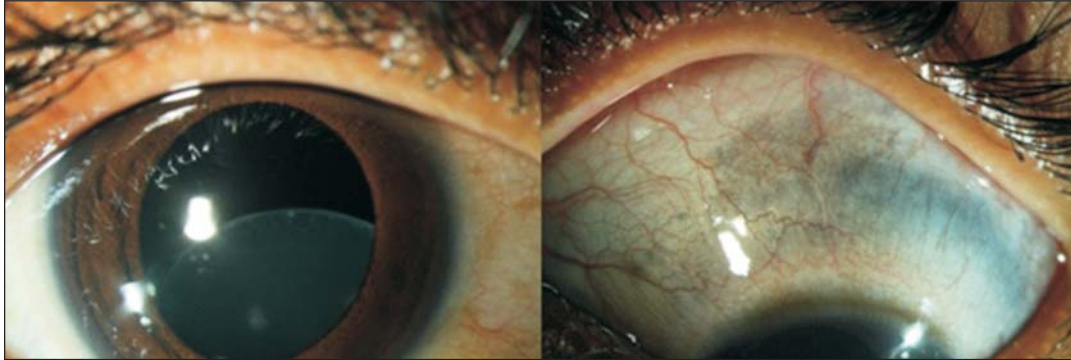


FIG 1a. Inferiorly subluxated crystalline lens

FIG 1b. Blue sclera without uveal ectasia in the left eye

An 8-year-old girl was detected to have corrected visual acuity of 6/18 in both eyes during a school screening programme. Both her eyes had inferiorly subluxated lenses, the superior border of the lenses lying centrally within the visual axis (Fig. 1a). There was an additional patch of blue discoloration of the sclera without uveal ectasia in the left eye (Fig. 1b). A diagnostic work-up for bilateral ectopia lentis revealed high serum homocysteine levels. Her parents were counselled for lensectomy on follow-up, advised a consultation with a paediatrician and initiation of pyridoxine therapy.

The ligaments responsible for keeping the lens in position and shape are abundant in cysteine, and deficiency of the same due to homocystinuria results in ectopia lentis. Blue sclera is typically associated with other causes of ectopia lentis affecting the collagen structure and is uncommon in homocystinuria. A simple torchlight examination of the eyes can therefore provide valuable clues towards a serious underlying metabolic disorder.

ANUBHA RATHI

BRIJESH TAKKAR

SHORYA AZAD

Dr Rajendra Prasad Centre for Ophthalmic Sciences

All India Institute of Medical Sciences

New Delhi

britak.aiims@gmail.com