

Case study

BLUXATED LENSES AFTER TRIVIAL TRAUMA

ABSTRACT:

Eye is a window to a rare disease. Two children aged 9 year old male and 5 years old female presented with pain, redness and sudden diminution of vision in right eye and left eye respectively following trivial trauma while playing. On complete ocular examination it was found that in the involved eye of both cases the lens has subluxated anteriorly causing pupillary block glaucoma. Bio-chemical findings include increased levels of homocysteine in blood- 52 μ mol/L in Case-1 and 26.2 μ mol/L in Case-2 (normal levels: 5.9-16 μ mol/L). Both children were not a known case of homocystenuria. They were first diagnosed as a case of homocystenuria by ophthalmologist.

KEY WORDS: homocystinuria, homocysteine, subluxation

INTRODUCTION:

Homocystinuria is a general term for a group of disorders that cause elevated levels of amino acid homocystine in urine. These disorders primarily occur because of deficiency of any of several enzymes required for the proper degradation or reformation of an essential amino acid called methionine. Elevated levels of methonine can cause a variety of health problems. There are seven biochemical types of homocystenurias which have been reported so far. The classical homocystinuria due to cystathionine beta-synthase (CBS) deficiency is an autosomal recessive disorder of methionine metabolism that produces increased levels of urinary homocysteine and methionine. Ocular manifestations include progressive ectopia lentis, detachment of retina and significant myopia. Defect in amino-acid metabolism results in brittle zonules that rupture. This allows the lens to displace inferonasally. There is no accommodation and condition may progress. It manifests itself in other organ systems such as vascular, central nervous system, and connective tissue. Skin manifestations include malar flush easily seen after vigorous exercise or after exposure to cold, livido reticularis on the extremities, and thinning and diffuse hypopigmentation of the hair, CNS manifestations include mental retardation, grand mal seizures, depression, chronic behavioral disorders, obsessive compulsive disorders, and personality disorders. Cardiovascular manifestations include thrombosis of both veins and arteries.^[1,2,3,4]

CASE REPORT:

Case-1 : A nine year old male child (Figure-1) presented with sudden diminution of vision in his right eye following trivial trauma while playing. Visual acuity in the right eye was hand

movements which did not improve with pin hole, while in the left eye was 2/60 which improved with pin hole to 6/12. Intraocular pressure on day of presentation was 60mmHg in right eye and 10mmHg in left eye which was measured with non contact tonometer (NCT). On slit lamp examination there was hyphema with anteriorly subluxated lens causing pupillary block glaucoma in the right eye, while in the left eye there was inferiorly subluxated lens (Figure-2). The child had typical associated features of homocystinuria i.e malar flush (Figure-3), thinning and diffuse hypopigmentation of hair, pes pectus (Figure-4), pes cavus (Figure-5), and mental retardation due to which he was not sent to any school for his education. He had myopia of >12D in his left eye. Refractive error could not be assessed in right eye. Bio-chemical findings include homocysteine in blood which was 52μmol/L (normal levels:5.9-16μmol/L). The child was not a known case of homocystinuria. Lensectomy was done under general anaesthesia with ACIOL implantation and his BCVA on followup after 3 months was 6/18. Parents were advised to give methionine-restricted, Cysteine-supplemented diets and pyridoxine in high doses (100 mg tid).

Case-2: A five year old female child(Figure-6) presented with sudden diminution of vision in her left eye following ocular trauma while playing. Visual acuity in the right eye was 3/60 which improved with pin hole to 6/18, while in the left eye was Finger counting close to face which did not improve with pin hole. Intraocular pressure on day of presentation was 11mmHg in right eye and 45mmHg in left eye which was measured with NCT. On slit lamp examination there was inferiorly subluxated lens in the right eye (Figure-7) and anteriorly subluxated lens causing pupillary block glaucoma in the left eye (Figure-8). The child did not have other associated features of homocystinuria except ectopia lentis and history of delayed milestones . She had myopia of >12D in her right eye. Refractive error could not be assessed in left eye. Bio-chemical findings include homocysteine in blood which was 26.2μmol/L(normal levels:5.9-16μmol/L). The child was not a known case of homocystinuria. This case represents an incomplete clinical form of the disease and may suffer from other associated features as she grows older if not managed in time by a pediatrician. Lensectomy was done without IOL and child was referred to higher institution for secondary IOL implantation. Pediatric consultation was taken for further management for preventing future complications. Parents were advised to give methionine-restricted, Cysteine-supplemented diets and pyridoxine in high doses (100 mg tid).

DISCUSSION:

In 1960, the first case of homocystinuria was reported from Northern Ireland^[5]. The patient was initially described as having an unusual case of Marfan syndrome with renal abnormalities at the age of 7 years. The patient was mentally slow and thin and had fair hair, pale skin and flushed cheeks. He had arachnodactyly, dolichostenomelia, pes cavus, a high arched palate, and bilaterally dislocated lenses. At age 10 years, the patients urine was found to contain a large quantity of homocysteine; urinalysis results for the nitroprusside cyanide test were positive.

Homocystinuria is caused by mutation in the CBS gene (21q22.3). It is inherited as autosomal recessive. The diagnosis of homocystinuria is often delayed. In a study which was done by Cruysberg et al^[6] there was a mean delay of 11 years between the first major signs of the disease and final diagnosis of homocystinuria. Lens dislocation is observed in 85% of the patients with homocystinuria. Myopia in excess of 5D is extremely rare in children under 5 years of age. If ocular fundus of a highly myopic patient shows no clear ophthalmoscopic signs of axial myopia and the corneal curvature is not abnormal, then lenticular myopia due to ectopia lentis should be suspected and searched for. So ophthalmologist may be first person to diagnose the case of homocystinuria.

Eye is a window to a rare disease. Both these cases, which we are reporting, were not a known case of homocystinuria. Clinical manifestations of homocystinuria vary markedly from severely affected to nearly normal. The clinical characteristics develop after birth and are progressive. In both our cases, a 11-year child had more clinical features than a five year old child. Ectopia lentis is a zonular disease where alteration of zonular fibres weakens the zonular support of lens which becomes vulnerable to dislocation with trivial trauma. In children presenting with the subluxated/dislocated lens after trivial trauma, detailed ocular examination of the uninvolved eye should be done. Since the children are operated under general anaesthesia measures should be taken to prevent development of thromboembolism and use of nitrous oxide should be avoided as it causes increase in the levels of blood homocystine by inhibiting methionine synthase. After surgical management both children were put on medical management. Parents were advised to give methionine-restricted, Cysteine-supplemented diets (Max.500mg/day) and pyridoxine in high doses (300-600mg/day). Folic acid and Vit.B₁₂ are used in partial responders. Betaine (3-methyl glycine) is used in pyridoxine resistant patients. Treatment is aimed at maintaining the plasma homocystine concentration below 11 µmol/L and the plasma homocysteine concentration as close to normal as possible. Serum homocysteine levels are done regularly to monitor response to treatment.^[7,8,9,10,11]

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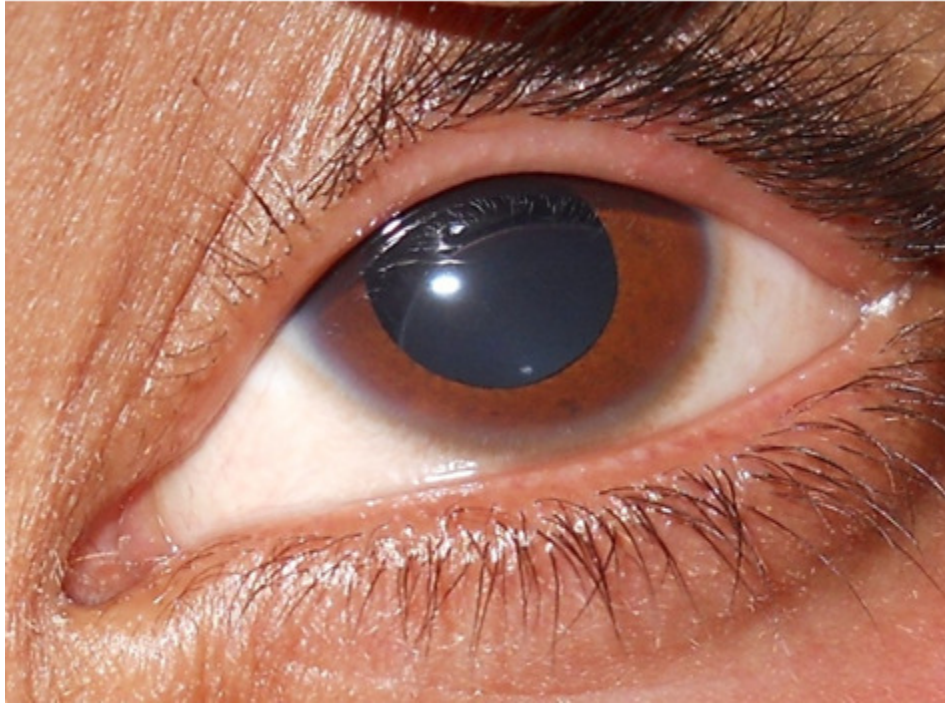
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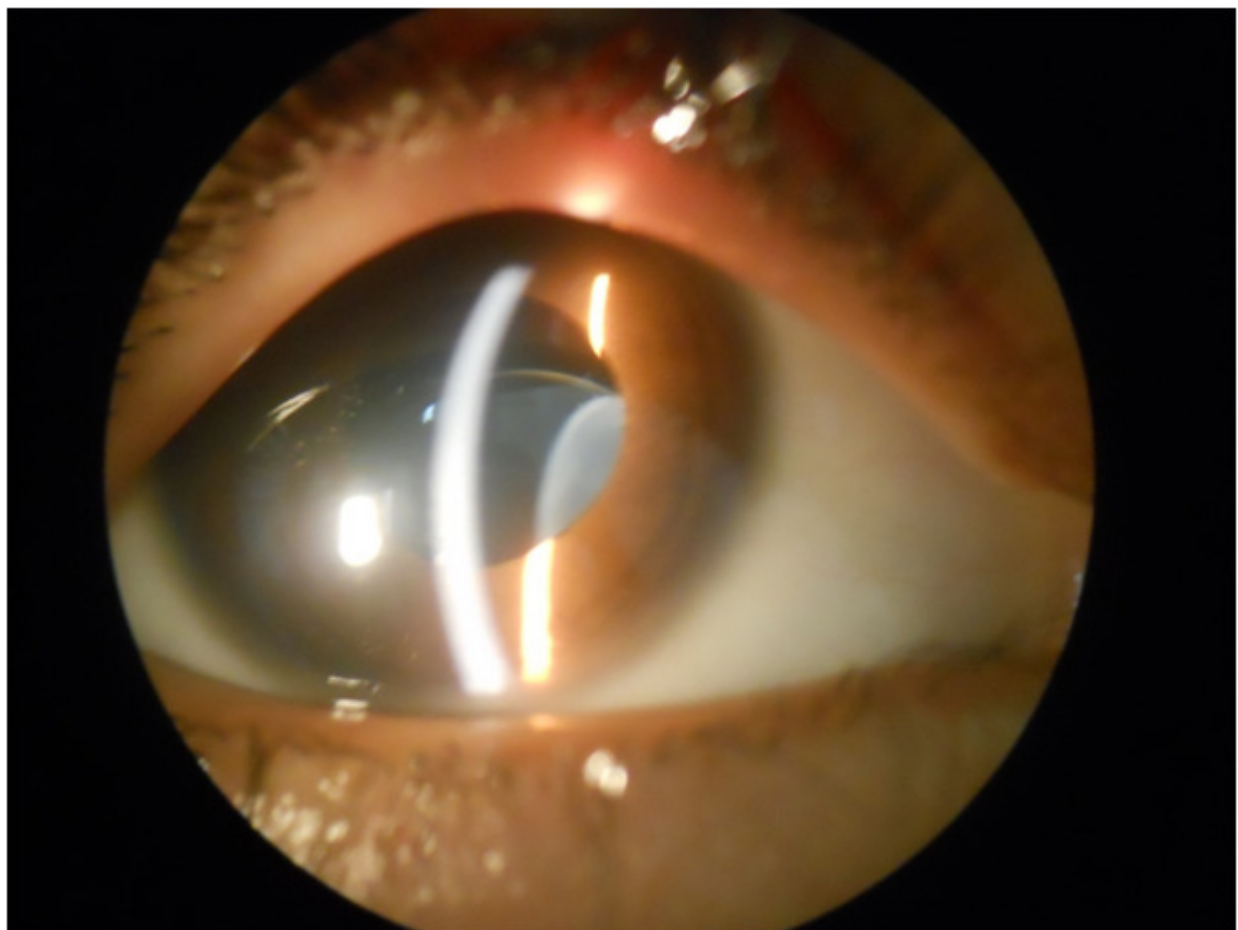
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133 FIGURE:5



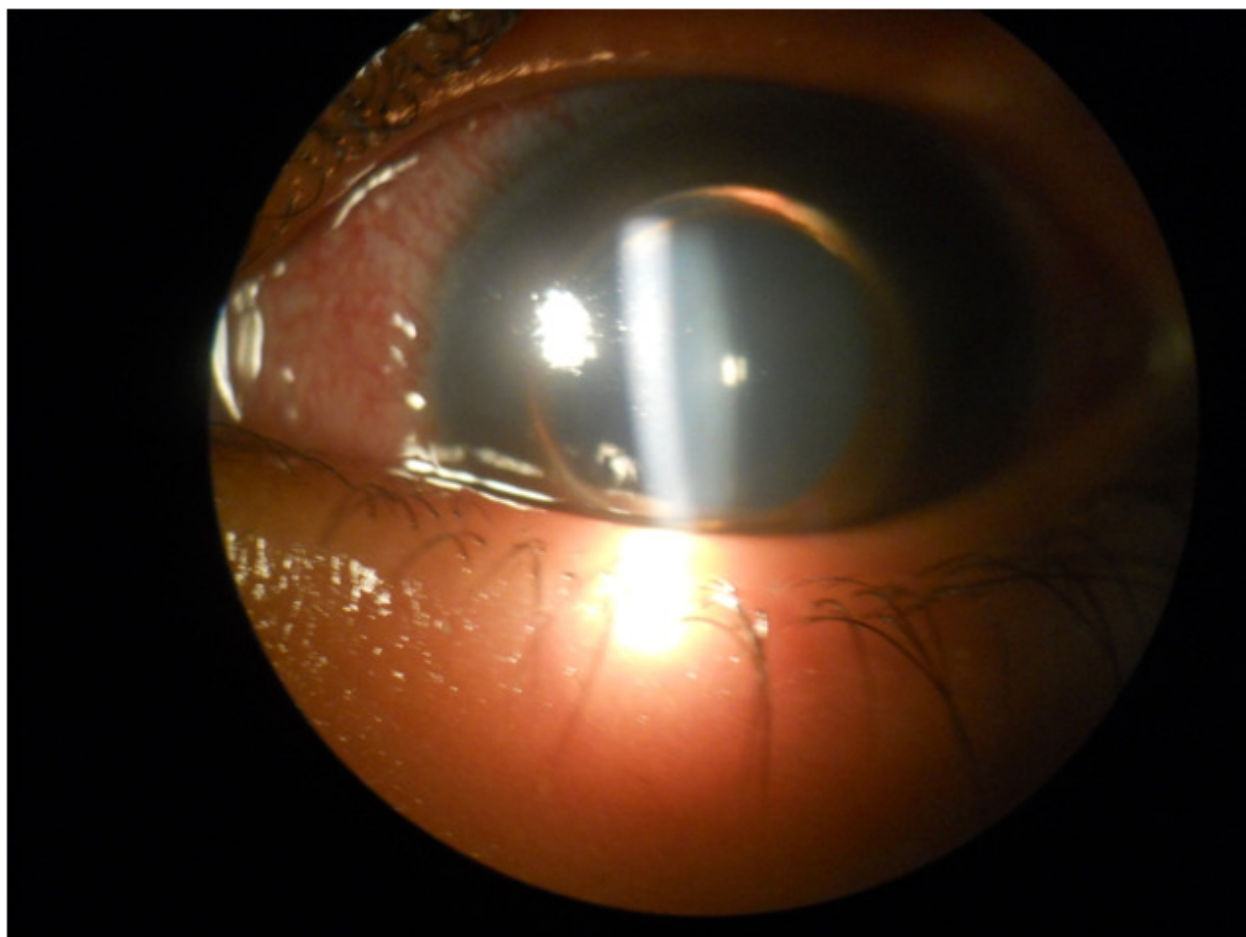
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FIGURE:8