



A CASE REPORT ON BILATERAL ECTOPIA LENTIS: A RARE CAUSE OF HOMOCYSTINURIA

Medicine

Surbhi Pareek

HOD, Deitetics Department, SDMH, Jaipur

Dr. Yadendra Gupta

Assistant Professor, Department Of Medicine Mahatma Gandhi Medical College, Jaipur.

Dr. Madhvi Sanwelka*

Senior Resident, Department Of Pathology, NIMS&R, Jaipur. *Corresponding Author

ABSTRACT

Serum homocysteine levels have been linked to eye diseases, such as venous and arterial occlusions, retinal emboli, glaucoma, and age-related macular degeneration. However, the link between homocysteine and lens dislocation has not been fully investigated. Thus, in this case report, we aimed to assess the association between lens dislocation and serum levels of homocysteine.

KEYWORDS

Homocystine, Ectopia lentis, Diet, dislocation

INTRODUCTION

Serum homocysteine levels have been linked to eye diseases, such as venous and arterial occlusions,¹ retinal emboli,² glaucoma,^{3,4} and age-related macular degeneration.⁵ However, the link between homocysteine and lens dislocation has not been fully investigated.⁶ Recently, it was hypothesized that homocysteine may be a lens epithelial stressor, which results in the development of unfolded protein aggregates in the lens.^{7,8} In human lens, there are epithelial cells whose prolonged exposure to homocysteine has been shown to induce unfolded protein response, resulting in lens dislocation.^{7,8} Homocystinuria (homocysteine > 100 μmol/L) is associated with congenital cataract^{9,10} and lens dislocation/luxation,^{9,10} but the association between higher serum homocysteine levels with lens dislocation is poorly understood.⁶ Vitamin B12 and folate are involved in homocysteine metabolism¹¹ and its use can lower homocysteine levels.

Thus, in this case report, we aimed to assess the association between lens dislocation and serum levels of homocysteine.

CASE REPORT

A 6 year-old child was brought by her parents history of decreased vision in both eyes. There was blurring and was found difficulty in reading on black board in school. There was no history of trauma or similar family history. On ocular examination, and 5+power were given in each eye. His eye power was increasing in every six months. Slit-lamp examination revealed bilateral inferonasal subluxation of crystalline lens, giving the appearance of 'setting sun' in the left eye. Ophthalmologist suggested surgery for lens dislocation. Fundus examination was unremarkable in both eyes. Laboratory investigation were done before surgery. His serum homocysteine levels were >50 μmol/l (reference range 4-15.3 μmol/l). he was operated on one eye and referred for further evaluation. Genetic test was done and patient was diagnosed with homocystinuria. These findings were consistent with diagnosis of bilateral ectopia lentis, secondary to classical homocystinuria. This patient was referred for diet modifications and diet counseling was started. Diet was planned with: Safe level of protein 0.91 gm/kg body weight Calories as per RDA. Blood homocystine level was again measured after two months and it was found to be decreased from >50 to 26 μmol/L. Again the same diet with Safe level of protein 0.91 gm/kg body weight Calories as per RDA was continued. After this report second eye got operated after six months of previous operation. Patient was examined by ophthalmologist for power in both eyes and was given +1 number in both eyes. Patient was discharged with diet restriction and advised to follow this restricted diet.

Patient came for follow-up after six months. lab investigations was done and found that homocystine levels was 6.39 μmol/L (Ref Range: 4-15.3 μmol/L).

DISCUSSION

Ectopia lentis is a rare ocular disorder associated with displacement of

crystalline lens in the eye.¹² It is a major ocular complication of homocystinuria, which is an autosomal recessive disorder of homocysteine metabolism and caused by deficiency of cystathionine β-synthase, leads to an abnormal accumulation of homocysteine and its metabolites in urine and blood.¹³ Abnormal concentration of glycoproteins in homocystinuria weakens the zonular fibres, causing subluxation of lens and increased curvature of lens with high myopia.¹⁴ Lens subluxation is most commonly found to be inferonasal. These patients are usually tall and thin, and 50% have developmental delay.¹⁵ Our patient underwent bilateral lensectomy with implantation of scleral-fixated intraocular lens. Her visual acuity in follow-up improved, with correction of refractive error also. Homocystinuria usually presents with multisystem involvement like skeletal, vascular, neurological and eye involvement. Early diagnosis in homocystinuria is essential to prevent intellectual disability and thromboembolic phenomena also.

Nutrients known to be associated with elevated homocysteine include vitamin B6, vitamin B12 and folate.¹⁴ It was suggested that adequate levels of vitamins are essential for homocysteine metabolism may be needed for optimal levels of homocysteine^{14,15} and that low levels of one or more of these vitamins might result in elevated levels of homocysteine.¹⁴

REFERENCES

- Chua B Kifley A Wong TY Homocysteine Mitchell P. and retinal vein occlusion: a population-based study. *Am J Ophthalmol.* 2005; 139: 181–182.
- Chua B Kifley A Wong TY Homocysteine Mitchell P. and retinal emboli: the Blue Mountains Eye Study. *Am J Ophthalmol.* 2006; 142: 322–324.
- Bleich S Roedl J von Ahnen N Elevated homocysteine levels in aqueous humor of patients with pseudoxfoliation glaucoma. *Am J Ophthalmol.* 2004; 138: 162–164.
- Roedl JB Bleich S Reulbach U Homocysteine levels in aqueous humor and plasma of patients with primary open-angle glaucoma. *J Neural Transm.* 2007; 114: 445–450.
- Rochtchina E Wang JJ Flood VM Mitchell P. Elevated serum homocysteine, low serum vitamin B12, folate, and age-related macular degeneration: the Blue Mountains Eye Study. *Am J Ophthalmol.* 2007; 143: 344–346.
- Sen SK Pukazhvanthen P Abraham R. Plasma homocysteine, folate and vitamin B12 levels in senile cataract. *Indian J Clin Biochem.* 2008; 23: 255–257.
- Shinohara T Ikesugi K Mulhern ML. Cataracts: role of the unfolded protein response. *Med Hypotheses.* 2006; 66: 365–370.
- Elanchezian R Palsamy P Madson CJ Lynch DW Shinohara T. Age-related cataracts: homocysteine coupled endoplasmic reticulum stress and suppression of Nrf2-dependent antioxidant protection. *Chem Biol Interact.* 2012; 200: 1–10.
- Presley GD Stinson IN Sidbury JB Jr. Ocular defects associated with homocystinuria. *South Med J.* 1969; 62: 944–946.
- Presley GD Sidbury JB. Homocystinuria and ocular defects. *Am J Ophthalmol.* 1967; 63: 1723–1727.
- Selhub J. Homocysteine metabolism. *Annu Rev Nutr.* 1999; 19: 217–246.
- Olitsky SE, Hug D, Plummer LS, Strass-Issern M. Abnormalities of the lens. In: Kliegman RM, Stanton BMD, St. Geme J, Scho NF, Behrman RE, eds. *Nelson Textbook of Pediatrics*. 19th edn. Philadelphia, Elsevier, 2011: 2171–2.
- Chen P, Poddar R, Tipa EV, Dibello PM, Moravec CD, Robinso K, et al. Homocysteine metabolism in cardiovascular cells and tissues: implications for hyperhomocysteinemia and cardiovascular disease. *Adv Enzyme Regul* 1999; 39: 93–109.
- Harrison D, Mullaney P, Mesfer S, Awad A, Dhindsa H. Management of ophthalmic complications of homocystinuria. *Ophthalmology* 1998; 105: 1886–90.
- Simon MA, Origlieri CA, Dinallo AM, Forbes BJ, Wagner RS, Guo S. New management strategies for ectopia lentis. *J Pediatr Ophthalmol Strabismus* 2015; 52: 269–81.