A rare metabolic case presenting to Ophthalmology

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Introduction

- Homocysteinuria is rare autosomal recessive disorder of methionine metabolism involving the transsulfuration or methylation pathway in methionine metabolism
- Incidence: 1 in 250,000
- Deficiencies of 3 enzymes namely methylene tetrahydrofolate reductase, methycobalamin synthase and cystathionin B-synthase lead to the accumulation of homocysteine and its metabolites
- Clinical features: marfanoid habitus, downward lens dislocation, myopia, seizures, flush on checks
- Main complication: thromboembolism, others include osteoporosis and short sightedness
- Treatment: low protein diet, Pyridoxine (vitamin B6), Betaine Vitamin B12 and risk management for thromboembolism

Case Report

- 25 years old female presented with rapid onset of loss of vision on both eyes
- Background history: well-controlled epilepsy, normal mental and physical development
- Family history: nothing of significant; no History of Marfan's Syndrome
- Examination: systemic review was unremarkable, clinically eu-thyroid and eu-adrenal. She had high arched palate with no other stigmata of Marfan's Syndrome
- Occular examination: Inferior lens dislocation in both eyes

Investigations

Laboratory Investigation

Routine Haematology – normal
Routine Biochemistry – normal
Vitamin B12 = 152mg/L (191-663)
Urine homocysteine = 524.5umol/L (2-14.2)
Plasma homocysteine = 237.9 & 254.5 umol/L(0-16)

Imaging

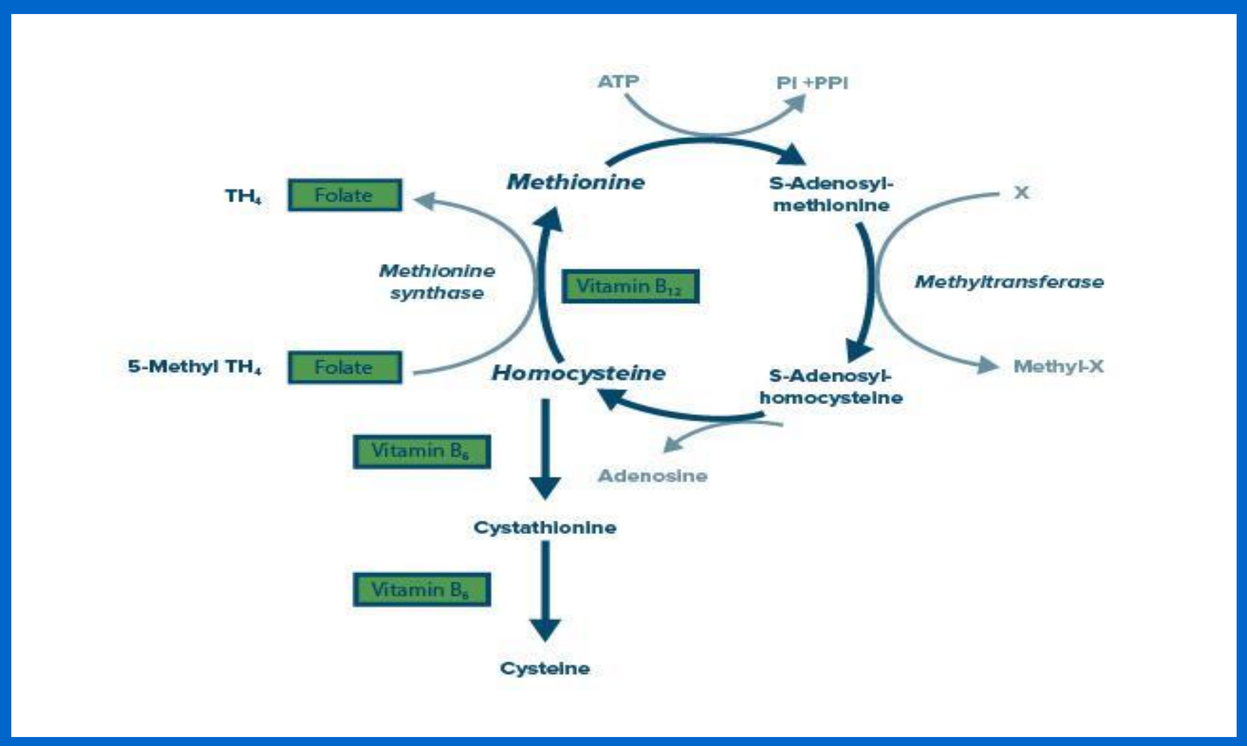
CT angiogram - normal aortic root dimension

Echo – normal

ECG - normal

Genetic Test results

Heterozygous pathogenic mutation on the Cystathionin B synthase (CBS) gene c.833T>C; p1278T.



Management

- Low protein diet
- Pyridoxine 100mg TDS (titrate to response)
- Folic acid supplementation
- Vitamin B12
- Referral to Ophthalmology for: Vitrectomy, lensectomy and right intra-occular lens implant
- Consider Betaine: promotes conversion of homocysteine back to methionine

Discussion

- Although visual disturbances have been described as a complication in thyroid eye disease, it can present in other metabolic conditions
- The risk of thromboembolism is increased in Homocysteinuria; therefore prompt diagnosis and treatment is essential especially in younger patients <30 years
- Female patient should be counselled for the increase risk of thromboembolism with pregnancy
- Rare metabolic conditions should be considered in patients with visual problems

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