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A CASE OF BILATERAL ECTOPIA LENTIS DUE TO HOMOCYSTINURIA

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Abstract : An 8year old boy presented with complaints of defective vision both eyes. On examination he was found to have dislocated lens in anterior chamber in right eye and inferior subluxation of lens in left eye . Since he was a known case of megaloblastic anemia, with history of herniotomy and features of ectopia lentis, investigations were done to confirm homocystinuria and was diagnosed to have homocystinuria. child underwent right eye lens removal with scleral fixated intra ocular lens implantataion. For left eye pars plana vitrectomy and lens removal with intraocular lens implantation is planned at a later date.

Keyword :ectopia lentis, homocystinuria, cystathionine synthase, pars plana vitrectomy

CASE REPORT:

An 8 year old male child presented with complaints defective vision both eyes for the past 3 years. There was no history of pain or redness of both eyes.

BIRTH HISTORY

Child was born through normal full term delivery, birth weight was 3kg

PAST HISTORY

child was on treatment for megaloblastic anaemia in children's hospital..He underwent right sided herniotomy surgery one year back.

FAMILY HISTORY

He is a third born child of second degree consanguineous marriage. He has two siblings, both of them were healthy. No history of similar illness in the family

ON GENERAL EXAMINATION:-

Child appears to be moderately built and nourished Pallor present No icterus or cyanosis or clubbing or generalised lymphadenopathy Vitals were stable

CVS,RS : normal

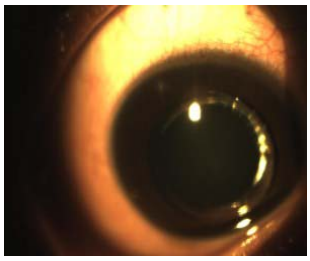
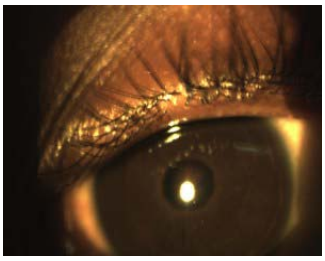
CNS: higher mental functions normal

Child is irritable and hyperactive with concentration difficulty. Intelligence appears to be normal.

ON OCULAR EXAMINATION:-

RIGHT EYE		LEFT EYE
2/60	VISION	3/60
12mmhg	TENSION(I care)	14mmhg
Normal	LIDS	Normal
Clear	CONJUNCTIVA	Clear
mild edema	CORNEA	Clear
Dislocated lens in anterior chamber	AC	Normal depth
Colour pattern normal	IRIS	Colour pattern normal
Details not made out	PUPIL	3mm, round, reacting to light
clear	LENS	Inferior subluxation of lens

RIGHT EYE LEFT EYE

FUNDUS:
Right eye- view hazy due to dislocated lens and corneal edema.
Lefteye- Media clear, disc and vessels normal
B SCAN:
Right eye- vitreous clear, retina attached, choroidal thickening present
Ultra sound biomicroscopy:
Left eye- zonular dialysis noted from 9 – 3 o'clock position with subluxation of lens and broad PAS from 12- 4'o clockposition
PROVISIONAL DIAGNOSIS:
BOTH EYES ECTOPIA LENTIS PROBABLY DUE TO HOMOCYSTINURIA. Since there was high suspicion of homocystinuria, we ordered for further investigations to confirm the diagnosis.
OTHER DETAILED INVESTIGATIONS:
Hb 9.8mg/dl
BT 1'50"
CT:3'20"
Serum homocysteine: 8.25 mic mol/L
URINE METABOLIC STUDY:
Cyanide nitroprusside test: positive
Benedict test ,Fecl3 test ,cetrimide test: Negative
Chest xray and USG abdomen: normal
CT spine: D4-D5 intervertebral disc prolapse
ECG& ECHO: normal
Cardiology opinion and paediatrician opinion were obtained no contraindication for surgical procedure.
DIAGNOSIS: BOTH EYES ECTOPIA LENTIS DUE TO HOMOCYSTINURIA..
TREATMENT:
Under general anesthesia, lens in right eye anterior chamber is removed and Scleral fixated intraocular lens implantation done.
POST OPERATIVE PERIOD:
Vision in Right eye: UCVA 6/18
Right eye -Anterior segment were normal. fundus examination -Cup disc ratio 0.3 ,normal with no evidence of glaucoma.
FURTHER PLAN:
Child is planned for Left eye Pars plana vitrectomy and lens removal with intra ocular lens implantation.
DISCUSSION:-
ECTOPIA LENTIS.:-
Next to cataract most common congenital anomaly of lens Mostly bilateral in congenital cases
Most common associations:
Marfan syndrome
Homocystinuria
Weill marchesani syndrome
Reiger s anomaly
Hyperlysemia
Ehler danlos syndrome
Sulphite oxidase deficiency.
HOMOCYSTINURIA:-
Second most common cause of bilateral subluxated lens
Autosomal recessive in nature Associated with mental retardation, positive biochemical test for urine homocystine Deficiency of more than one enzyme in methionine metabolism Mostly due to lack of enzyme **cystathionine synthase** Excess of methionine and homocystine in blood
OCULAR FEATURES OF HOMOCYSTINURIA:
Bilateral progressive subluxation of lens
Tremulous iris
Thin sclera
Ambylopia
Squint
Glaucoma
OTHER FEATURES:
Limbs are long
Arachnodactyly
Flat feet
Floppy gait
Hernia most common due to abdominal muscle under development
Spine – kyphosis scoliosis
Joints are prone for subluxation
Congenital anomalies of heart Thromboembolic events and CVA can occur in early adult hood Low IQ
INVESTIGATIONS:
Cyanide sodium nitroprusside test is a good screening test.
MANAGEMENT:
Diet restricted of methionine and supplementaion of oral cystine High doses of pyridoxine 600-1200mg daily and folic acid orally.
CONCLUSION:
All cases of homocystinuria ,as soon as diagnosed ,should be screened for ophthalmology related problems. Early diagnosis and intervention saves the child from ambylopia.
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