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CASE REPORT

BILATERAL CONGENITAL GLAUCOMA IN NEUROFIBROMATOSIS TYPE 1 AND MANAGEMENT (A RARE - CASE REPORT)

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ABSTRACT

Neurofibromatosis 1 is a genetic disorder of neural crest-derived cells. Riccardi classified NF into 8 subtypes NF-I to NF-VIII. NF-V is segmental NF (Prevalence 0.0014 and 0.002%) Caused by a Postzygotic somatic mutation, Café-au-lait macules & neurofibromas in a single, unilateral segment of the body, no family history, no systemic involvement. SNF classified into true segmental & Bilateral SNF. Glaucoma is a rare manifestation ofNeurofibromatosis -1. Patients usually have cafeaulait spots, plexiform neurofibroma of upper lid, and axillary freckles. If plexiform neurofibroma is present a 50% chance of developing glaucoma exists. Glaucoma is always unilateral, usually exists at birth or shortly after birth. We present a case of bilateral congenital glaucoma in Neurofibromatosis type 1 and management.

METHODOLOGY:

Diagnostic criteria Neurofibromatosis 1 least two of the seven

1) Six or more café-au-lait macules over 5 mm in greatest diameter in pre-pubertal individuals and over 15 mm in greatest diameter in post-pubertal individuals,

(2) Two or more neurofibromas of any type or one plexiform neurofibroma,

(3) Freckling in the axillary or inguinal regions,

(4) Optic glioma,

(5) Two or more Lisch nodules,

(6) Any distinctive osseous lesion such as sphenoid dysplasia or thinning of the long bone cortex with or without pseudarthrosis.

(7) A first degree relative with NF-1.

RESULTS:

HISTORY: A 4 day Female baby was presented by its mother with bilateral cloudy cornea's, enlarged eye balls noticed by parents. Multiple lesions on face and whole body, enlarged eyelids and watering in both eyes

Family H / O 1st child Non Consanginous Marriage, no similar history in other family members

Birth H / O FTND Birth Wt 2.6 Kg cried immediately, NO post natal complications.

Figure 1:

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GENERAL PHYSICAL EXAMINATION:

Multiple cafe lieu spots > 8 in number 0.5 mm in size over the trunk and extremities

Vital signs Stable, No other significant abnormality detected.

SYSTEMIC EXAMINATION by Neonatologist: NORMAL.

INVESTIGATIONS:

Blood, X ray chest, USG, Neurosonogram, ALL INVESTIGATIONS with in NORMAL limits.

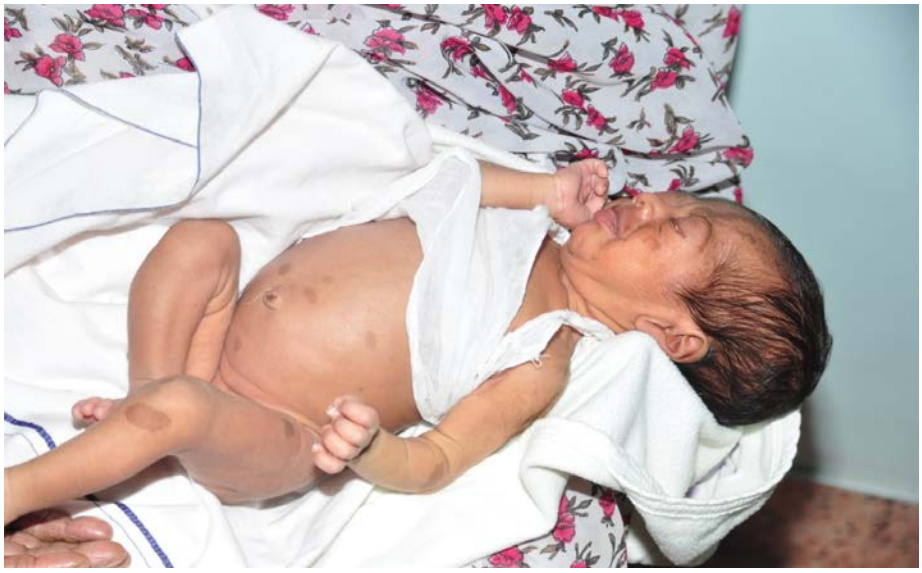

Figure 2:

Table 1:

	Right	Left
Lids	Plexiform neurofibroma	Plexiform neurofibroma
Conjunctiva	Mild congetion	Mild congetion
Cornea	Megalocornea	Megalocornea
	Hazy odema	< Hazy odema
	14 mm size	14 mm size
A C	Deep	Deep
Pupil	Reacting	Reacting
Lens	Clear	Clear
IOP	25 mm Hg	30 mm Hg

Figure 3:

Figure 4:

MANAGEMENT:

Both Eyes: Trabeculectomy with Trabeculotomy under General Anesthesia was done. A routine Trabeculectomy with Trabeculotomy under General Anesthesia was done with Mytomycin-C used for 4 min, a 4 by 3 mm triangular Flap raised, both sides Trabecolotomy was done, trebacular meshwork was punched, PI was done, Apical suture only applied 10-0 nylon, conjunctiva closed with vicryl 8-0. Post Operative Care involved Antibiotic Steroid Eye-Oint Bid x 8 wks, Regular 3-4 month follow up, IOP well controlled in low Teens.

DISSCUSSION:

Patients usually have cafeaulait spots, plexiform neurofibroma of upper lid, and axillary freckles. If plexiform neurofibroma is present a 50% chance of developing glaucoma exists. Glaucoma is always unilateral, usually exists at birth or shortly after birth. Our case has presented with all diagnostic features of neurofibromatosis 1, but having bilateral plexiform neurofibromas and coexisting glaucoma in both eyes which is a rare presentention very few times reported in the past. The patient was successfully treated surgically with regular follow ups.

CONCLUSION:

Our case is rare & classical anatomical presentation of Riccardi classified NF V subtype, Bilateral Segmental Neurofibromatosis with Bilateral Congenital Glaucoma with No family H/O No systemic involvement.

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