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Spectrum of Etiology for Children Presenting With Macrocephaly in Tertiary Care Hospital

Dr Mohammed Arif, RajibChatterjee, DrHimanshu S raichandani
PG candidate for MD pediatrics,
Professor and HOU,PG guide ,MBBS,DCH,MD pediatrics
PG candidate for MD Pediatrics

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ABSTRACT---
Macrocephaly is the condition in which the head circumference of an infant is above 2 standard deviations, which is above the 97th percentile. It can be due to benign conditions or can be due to underlying causes. Macrocephaly is found in about 2% to 3% of the population. There is not a significant difference between genders. The pathogenesis of macrocephaly is etiology-specific as it can result from overgrowth of the skull bones or an increase in the volume of the intracranial structures like CSF, blood, or the brain parenchyma. . The management of macrocephaly is dependent upon the exact etiology.,The prognosis of macrocephaly is determined by the underlying cause

Conclusion: Macrocephaly is a life threatening yet treatable condition which if ignored may results in long term consequences and sequelae. Preventable cause like ingestion of folic acid during pregnancy, ANC scan during pregnancy to look for any anomaly in baby, taking iron tablets in thalassemia child, VP shunt operation in hydrocephalus, child care, proper family environment may help preventing the condition. On the other hand early detection of congenital malformation, proper physical and laboratory examination may help detect the underlying cause and treat accordingly.

Keywords—macrocephaly, hydrocephalus, porencephaly, thalassemia major, hemangioma,

I. INTRODUCTION

Macrocephaly is defined as an occipitofrontal circumference greater than two standard deviation (SD) above the mean for age and sex. Megalencephaly or enlargement of the brain parenchyma may be familial or associated with inherited syndromes or neurometabolic disease. Infants with benign familial megalencephaly have increase head size at birth that persists through infancy along the upper growth curve percentile, and is associated with normal body size, neurologic examination and development. Children with metabolic causes have normal head circumference at birth; macrocephaly is noted as the child gets older. Diagnosis is suggested by accompanying features and biochemical abnormalitites.

Hydrocephalus,characterised by an excessive amount of CSF, may be caused by increased production, decreased absorption, or obstruction to flow. Most patients show postnatal rapid increase in head size and are symptomatic due to underlying disease or raised

Case series

Case 1

G3A3L1 preterm 36 weeks born by vaginal delivery severe oligohydromnios VLBW RDS grade 1 with hydrocephalus with symmetrical IUGR with birth weight of 1.1 kg,BBG—B+
DOB—23/7/2017
TOB—10.22 AM
Operated on 24/1/2020

Head circumference	Age
30 cm	Birth
32.1 cm	At 7th day of life
33 cm	At 14th day of life
33.8 cm	At 21st day of life
35 cm	4th month
36.5 cm	5th month
39.5 cm	7th month
42 cm	10th month
54 cm	1 year 8 months
55 cm	1 year 10 months

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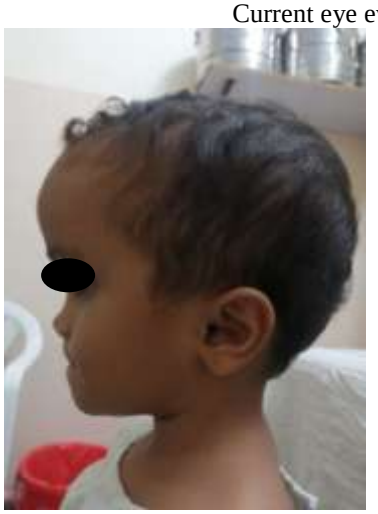
56 cm	3 years 10 months

NSG – all ventricles appear dilated S/O communicating hydrocephalus

	Right	**Left**
Lateral ventricle		
1) Frontal horn	44mm	45mm
2) Temporal horn	21mm	22mm
3) Occipital horn	44mm	45mm
Third ventricle	22mm	
Fourth ventricle	34mm	

USG abdo pelvis—normal
Lumbar puncture done at birth – 2 cells ,all lymphocytes , glucose—20mg/dl (parallel blood glucose—60 mg/dl),Protein—124mg/dl, LDH—127 IU/l
2D echo--- normal
Chest Xray—normal

CT brain plain—Grossly dilated bilateral lateral,third,fourthvenricle without obvious visualised intra or extra luminal occlusive cause
MRI brain plain—dilated ventricle with periventricular hypodensity,thinning of overlying cerebral cortex and corpus callosum,choroidal cyst S/O increased ICT.
ROP at birth—no evidence of papilodema
Current eye evaluation—secondary optic atrophy



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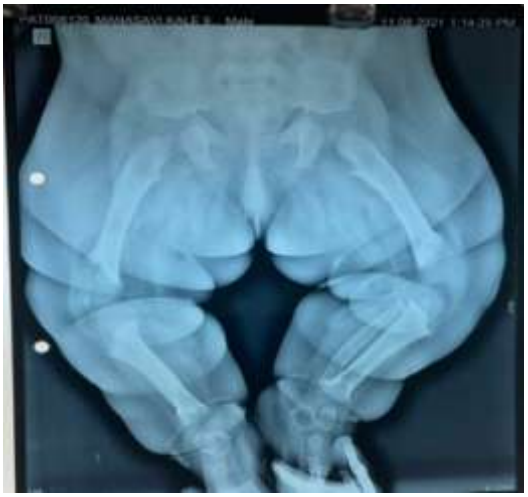
Case 2

Full term male LSCS i/v/o short stature of mother, CIAB (cried immendiately after birth) with birth weight of 2.1 kg and head circumference of 31 cm, no NICU stay came to us with complain of not gaining height and large head.

On examination



Developmentaly child is normal in all 4 domains

Child is immunized as per NIP

At 1 year of age head circumference is 49 cm. and diagnosed as a case of short stature and achondroplasia and macrocephaly.

Case 3

Full term male vertex vaginal delivery CIAB with birth weight of 2.5 kg, head circumference of 32 cm at birth 3rd degree consanguineous marriage came to us at 2 years 9 months with complain of not able to stand and seizure episode GTCS type (febrile seizure). Global developmental delay seen. Grade 4 reflex in lower limbs and grade 2 in upper limbs with ankle clonus +.

On examination

Flat occiput

Frontal bossing

Coarse face

Macrocephaly

Short neck

Head circumference at 2 years 9 months is 53 cm.

And diagnosed as a case of neuroregression with dysmorphicfacies with macrocephaly.

Case 4

DOL 11, Singleton, Term (38 weeks POG), Male, 1st by birth order of nonconsanguineous marriage admitted i/v/o Low Birth Weight and came to us for Review Neurosonography with head circumference at birth was 50 cm.

Birth weght-2 KG

Disproportionate short stature

Macrocephaly with frontal bossing

Depressed nasal bridge

Rhizomelic shortening of arms with redundant skin folds on limbs

Brachydactyly

Weight on admission: 2.3 kg

MBG: O Positive

BBG: B Positive DCT: Negative

DOB:3/5/2022

TOB:7.20 PM

Birth history: Term/Full term normal vaginal delivery/ Male /SGA/ Cried immediately after bith/No H/o NICU stay at birth

Neurosonography at birth s/o Grade 1 Germinal Matrix Hemorrhage

O/E

Baby pale and sluggish Activity.No seizure episodes .Tolerating Breastfeeds well. Urine and stool passed .Anterior Fontanelle: widely open and bulging. Sagittal suture: widely separated upto glabella

CHEST XRAY (8/6/22): Normal

USG ABDO PELVIS (8/6/22): Normal

Neurosonography (8/6/22) s/o Grade 4 Germinal Matrix Hemorrhage and Cystic Encephalomalacia

MRI BRAIN(9/6/22): Subacute hemorrhage involving thalamus and midbrain on right side with intraventricular extension into right lateral ventricle with compression of 3 rd ventricle with resultant dilation of bilateral lateral ventricles. Extensive cystic encephalomalacia near completely replacing the brain parenchyma in right frontoparieto temporal and left frontal lobes, communicating with right lateral ventricle and causing mass effect in the form of midline shift of 10mm towards left side and diagnosed as a case of porencephaly with macrocephaly.

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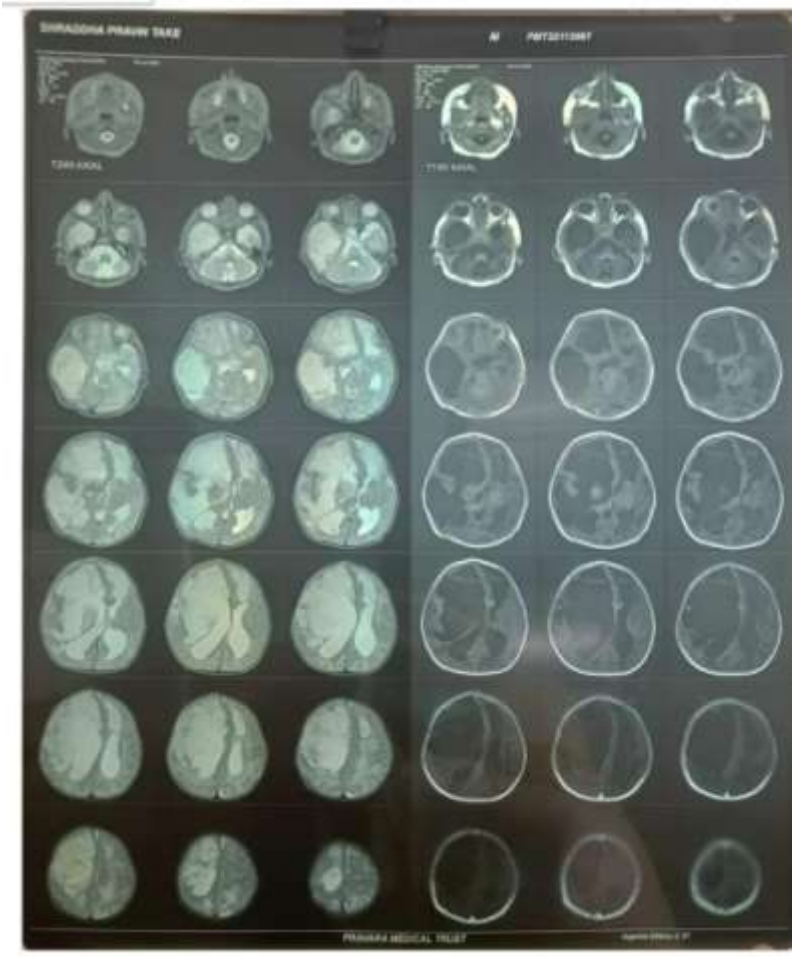

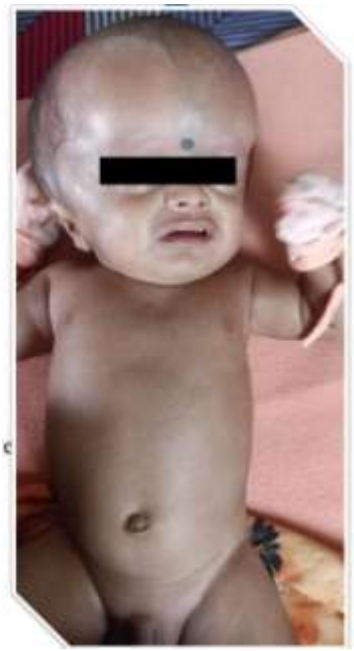

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Case 5

Full term normal vaginal delivery female , CIAB (cried immendiately after birth) with birth weight of 2.4 kg and head circumference of 40 cm, NICU stay for 5 days came to us with complain of not gaining height and large head.
On examination
Disproportionate short stature
Macrocephaly
frontal bossing
Rhizomelic shortening of arms with redundant skin folds on limbs
Genu varum
Developmentaly child is normal in all 4 domains
Child is immunized as per NIP
At 2 year of age head circumference is 50 cm. and diagnosed as a case of short stature and achondroplasia with macrocephaly.

Case 6

Full term normal vaginal delivery male child with birth weight of 2.6 kg delivered in PMT LONI,head circumference at birth is 30 cm came to us frequently for blood transfusion and is a known case of thalassemia major,head circumference at 5 years of age is 54 cm. delevopmental history is normal and child is immunized as per NIP.
On examination
Severe pallor
Jaundice
Frontal bossing
Malar eminence
USG abdomen and pelvis—hepatosplenomegaly
And diagnosed as a case of B thalassemia major with macrocephaly.

Case 7

Full term female LSCS i/v/o obstructed labour birth weight of 3.1 kg delivered in PMT lonion 22/10/2021 ,CIAB admitted to NICU with large head and meningomyelocele ,head circumference at birth was 52 cm . Mother has not taken tablet folic acid during pregnancy.
ANC scan suggestive of neural tube defect,compressed cerebellum, dilated bilateral ventricles suggestive of Arnold Chairy malformation type 2 with polyhydroamnios .
On examination
wide open AF
Head circumference more than 97th percentile
Eyes show sunset sign
Lumbosacral region had defect s/o Myelomeningocele with CSF leak
Lower limb hypotonia
Transillumination test positive
Dilated scapular veins.
NSG --- Severe dilation of ventricles compressing brain parenchyma s/o gross hydrocephalus.
Parents took DAMA (discharge against medical advice) and baby got expired after 3 days of discharge at home.

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Case 8

6 years old male child came to us with complain of paleness of skin and is a known case of B thalassemia major. Patient is a diagnosed case at 7th months of life and from then he is taking repeated blood transfusion. Patient is not taking tablet deferasirox regularly.

On examination

Frontal bossing
Malar prominence
Open mouth
Severe pallor
Growth retardation
Hepatosplenomegaly
Dark urine
Depressed nasal bridge

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Case 9

Baby came to us at 5th day of life 37 weeks/female/4th by order of non consanguineous marriage was born by normal vaginal delivery and brought to us with c/o
respiratory distress since 2 days
fever since 2 days
reduced oral intake
fever was of high grade and decreased by taking medication

O/E
Baby is icteric(krammer zone 4)
Lethargic
AF is open and wide
Lower intercostal retraction +
Tachypnea + on admission
Urine and stool passed
Later child was diagnosed as a case of meningitis on CSF examination and on follow up head circumference was more than 97th percentile.

	1st time	**2nd**	**3rd**	**`4th`**	**5th**
Cells	2	400	500	60	2
Leucocytes	0 %	40%	70%	40%	0 %
Lymphocytes	100 %	60%	30%	60%	100%
Glucose	32	50	54	72	80
Protein	72	78	70	40	34

Case 10

Full term normal vaginal delivery CIAB,with birth weight of 2.5 kg and birth head circumference of 30 cm came to us at 8th month of life with complain of fever,and multiple episodes of uprolling of eyes and tightening of all limbs. Developmental history is normal in all 4 domains.
On examination capillary hemangioma seen over anterior fontanelle.

Head circumference – 47 cm (macrocephaly)
USG local swelling was done- heterogenous round to oval lesion over anterior fontanelle showing excessive vascularity on Doppler study with feeding and draining vessels S/O hemangioma.
Ophthalmic examination- no sign of papilledema seen

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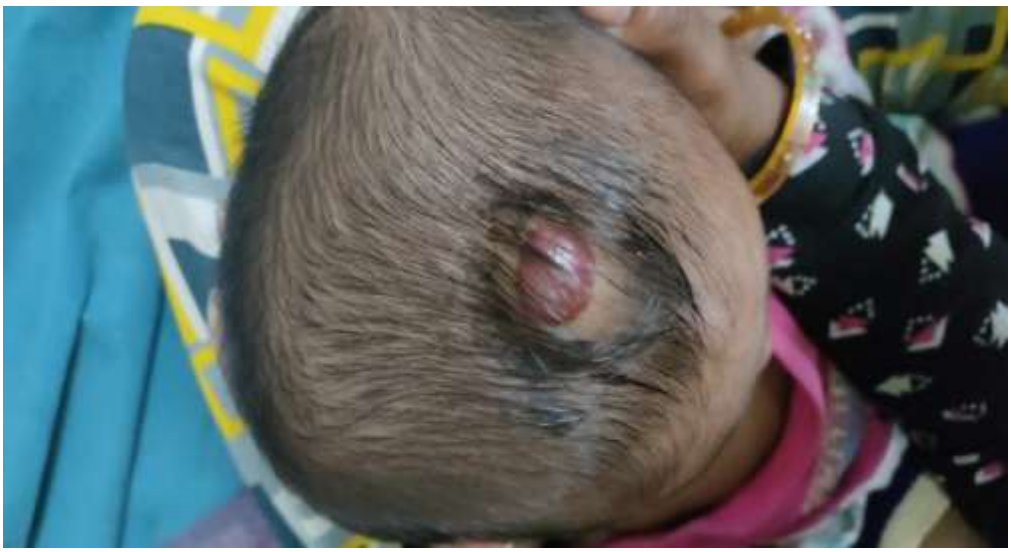
CSF examination

	On admission	Later after treatment
Cells	50	28
Lymphocytes	90%	100%
Leucocytes	10%	0 %
Gluocose	64 (parallel BSL—106)	47 (parallel BSl-- 90)
Protein	73	99



II. DISCUSSION

Evaluation of macrocephaly is indicated, if the head circumference is above 3 SD of the mean of the age and sex,or when serial measurements reveal progressive enlargement, as suggested by an increase by >2 cm per month during first 6 months of life, or the crossing of one or more major percentile lines between routine visits. Measurement of head size in parents is useful in diagnosing familial cases. Majority of patient requirecranial imaging,ultrasonography or CT scan.

III. CONCLUSION

Macrocephaly is a life threatening yet treatable condition which if ignored may results in long term consequences and sequelae. Preventable cause like ingestion of folic acid during pregnancy ,ANC scan during pregnancy to look for any anomaly in baby, taking iron tablets in thalassemia child, VP shunt operation in hydrocephalus ,child care, proper family environment may help preventing the condition. On the other hand early detection of congenital malformation, proper physical and laboratory examination may help detect the underlying cause and treat accordingly.

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