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

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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 result 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

<table>
<thead>
<tr>
<th>Head circumference</th>
<th>Age</th>
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<tbody>
<tr>
<td>30 cm</td>
<td>Birth</td>
</tr>
<tr>
<td>32.1 cm</td>
<td>At 7th day of life</td>
</tr>
<tr>
<td>33 cm</td>
<td>At 14th day of life</td>
</tr>
<tr>
<td>33.8 cm</td>
<td>At 21st day of life</td>
</tr>
<tr>
<td>35 cm</td>
<td>4th month</td>
</tr>
<tr>
<td>36.5 cm</td>
<td>5th month</td>
</tr>
<tr>
<td>39.5 cm</td>
<td>7th month</td>
</tr>
<tr>
<td>42 cm</td>
<td>10th month</td>
</tr>
<tr>
<td>54 cm</td>
<td>1 year 8 months</td>
</tr>
<tr>
<td>55 cm</td>
<td>1 year 10 months</td>
</tr>
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</table>
NSG – all ventricles appear dilated S/O communicating hydrocephalus

<table>
<thead>
<tr>
<th></th>
<th>Right</th>
<th>Left</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lateral ventricle</td>
<td></td>
<td></td>
</tr>
<tr>
<td>1) Frontal horn</td>
<td>44mm</td>
<td>45mm</td>
</tr>
<tr>
<td>2) Temporal horn</td>
<td>21mm</td>
<td>22mm</td>
</tr>
<tr>
<td>3) Occipital horn</td>
<td>44mm</td>
<td>45mm</td>
</tr>
<tr>
<td>Third ventricle</td>
<td>22mm</td>
<td></td>
</tr>
<tr>
<td>Fourth ventricle</td>
<td>34mm</td>
<td></td>
</tr>
</tbody>
</table>

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, fourth ventricle 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
Case 2
Full term male LSCS i/v/o short stature of mother, CIAB (cried immediately 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
Disproportionate short stature
Macrocephaly with frontal bossing
Depressed nasal bridge
Rhizomelic shortening of arms with redundant skin folds on limbs
Brachydactyly

Developmental 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 weight-2 KG
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 birth/No H/o NICU stay at birth
Neurosonography at birth s/o Grade 1 Germinal Matrix Hemorrhage
O/E
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.
Case 5
Full term normal vaginal delivery female, CIAB (cried immediately 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
Developmental 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. Developmental 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 LonI on 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.
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
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.

<table>
<thead>
<tr>
<th></th>
<th>1st time</th>
<th>2nd</th>
<th>3rd</th>
<th>4th</th>
<th>5th</th>
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</thead>
<tbody>
<tr>
<td>Cells</td>
<td>2</td>
<td>400</td>
<td>500</td>
<td>60</td>
<td>2</td>
</tr>
<tr>
<td>Leucocytes</td>
<td>0 %</td>
<td>40%</td>
<td>70%</td>
<td>40%</td>
<td>0%</td>
</tr>
<tr>
<td>Lymphocytes</td>
<td>100 %</td>
<td>60%</td>
<td>30%</td>
<td>60%</td>
<td>100%</td>
</tr>
<tr>
<td>Glucose</td>
<td>32</td>
<td>50</td>
<td>54</td>
<td>72</td>
<td>80</td>
</tr>
<tr>
<td>Protein</td>
<td>72</td>
<td>78</td>
<td>70</td>
<td>40</td>
<td>34</td>
</tr>
</tbody>
</table>

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
CSF examination

<table>
<thead>
<tr>
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<th>On admission</th>
<th>Later after treatment</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cells</td>
<td>50</td>
<td>28</td>
</tr>
<tr>
<td>Lymphocytes</td>
<td>90%</td>
<td>100%</td>
</tr>
<tr>
<td>Leucocytes</td>
<td>10%</td>
<td>0%</td>
</tr>
<tr>
<td>Glucose</td>
<td>64 (parallel BSL—106)</td>
<td>47 (parallel BSL—90)</td>
</tr>
<tr>
<td>Protein</td>
<td>73</td>
<td>99</td>
</tr>
</tbody>
</table>

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 require cranial 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.

REFERENCE