

Craniosynostosis

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Abstract

Craniosynostosis is a condition of one or more of the fibrous sutures in an infant skull prematurely fuses by turning into bone (ossification).¹ Premature fusion of cranial sutures may have an effect on cranial shape and less frequently on brain growth.² There is a changing in the growth pattern of the skull and the skull can't expand perpendicular to the fused suture, craniosynostosis can lead to abnormal head shape and abnormal facial features and results in increased intracranial pressure leading possibly to visual difficulty, sleeping impairment, eating problem, or impaired mental status with a significant reduction in IQ.

Keywords: Preterm labor, IUGR, Hydrocephalus, Fetal anomalies, Increase intracranial pressure.

Introduction

Craniosynostosis is relatively common developmental anomaly affects 1 in 2000 children and this is because the normal development of the skull is disrupted. In between the plates of bone's narrow openings are called sutures. The sutures give the bone plates flexibility so that the skull is able to grow along with the brain. This is important because the human brain grows rapidly following birth, literally doubling in size during the first three years of life, and the skull must grow to accommodate this growth.

After the skull and brain have grown to their full adult size, the sutures fuse together to create a single structure of bone. However, in cases of craniosynostosis, one or more sutures fuse together before birth or shortly afterwards.³ Potential risk factors include white maternal race, advanced maternal age, male infant sex,⁴ maternal smoking,⁵ maternal residence at high altitude,⁶ use of nitrosatable drugs in first trimester (e.g., nitrofurantoin, chlorthalidone, chlorpheniramine).⁷

Physical findings can include calvarial dysmorphism, midface hypoplasia, hydrocephalus, deafness, blindness, mental retardation, and extremity anomalies. The diagnosis, management, and treatment of craniosynostosis can be complex. Interdisciplinary team care has been practiced for many years in the care of children with cleft lip and cleft palate and complex. Craniosynostosis can either be:

1. **Syndromic:** Where the condition is one of a number of birth defects to affect a child
2. **Nonsyndromic:** Where the condition develops in isolation and the child has no other birth defects⁸

Nonsyndromic craniosynostosis is the most common form of the condition, accounting for 80%-95% of all cases. The cause of nonsyndromic craniosynostosis is unknown.

There are more than 150 different syndromes that can cause syndromic craniosynostosis, all of which are

very rare. A syndrome describes a range of different symptoms that are all related to a common cause, which is usually (but not always) genetic.

Craniosynostosis is very heterogeneous in terms of its causes, presentation, and management. Both environmental factors and genetic factors are associated with development of craniosynostosis. FGFR2, FGFR3, FGFR1, TWIST1 and EFNB1 genes are major causative genes of genetic syndromes associated with craniosynostosis. Although most of syndromic craniosynostosis show autosomal dominant inheritance, approximately half of patients are de novo cases. Apert syndrome, Pfeiffer syndrome, Crouzon syndrome, and Antley-Bixler syndrome are related to mutations in FGFR family (especially in FGFR2), and mutations in FGFR3 can be overlapped between different syndromes. Saethre-Chotzen syndrome, Muenke syndrome, and craniofrontonasal syndrome are representative disorders showing isolated coronal suture involvement. Compared to the other types of craniosynostosis, single gene mutations can be more frequently detected, in one-third of coronal synostosis patients. Molecular diagnosis can be helpful to provide adequate genetic counseling and guidance for patients with syndromic craniosynostosis.⁹ Fibroblast growth factor and fibroblast growth factor receptor (FGFR) regulate fetal osteogenic growth and are expressed in cranial sutures in early fetal life. These factors possibly influence fetal suture patency. Mutations in the gene coding for FGFR1 cause Pfeiffer's disease, and mutations in FGFR2 cause Apert's syndrome and Crouzon's disease.⁷

Case Report

A 30yrs old primi gravida female came to opd for routine antenatal check up. No h/o consanguineous marriage. Her last menstrual period date was (07/01/2017). 3yrs of active marriage life. 70kg weight and BMI -34.6.

On Examination: Blood pressure -130/80 mmHg, Pulse rate -90/bpm, respiratory rate -16/min, SPO₂-98% & per abdominally 28 to 30 weeks size of uterus with regular FHS on auscultation.

USG Findings & Doppler Study: fetal heart rate -139 BPM, fetal movement normal, cephalic presentation with lemon shaped skull, placenta posterior grade 1 maturity, amniotic fluid index-14, EFW-1311gm, cervical length -35mm, cervical os closed. On Doppler examination Normal flow pattern in both uterine arteries and umbilical artery and middle cerebral artery, normal findings of all 4 cardiac chambers.

Investigation: Hb - 9.10gm%, WBC-10800cumm, Platelets-379000cumm, BGRH - AB positive, HIV-NR, HB_sAG- NR, RBS-108mg/dl, Urea-24.2mg/dl, Creatinine-0.69mg/dl.

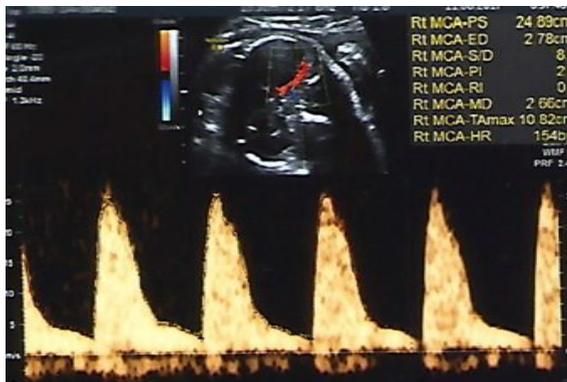


Fig. 1: MCA Doppler of fetus



Fig. 2: Fetus in lemon shape skull



Fig. 3: Depressed ant. frontanalle fetus



Fig. 4: Hydrocephalus in patient



Fig. 5: Full usg analysis

Management: A 30 Years old primi gravida female came to opd for routine antenatal check up. As per usg finding she diagnosed as a lemon shaped skull and hydrocephalus (Fig. 1,2,3,4,5). usg report convey to patient and advised for further evaluation (karyo typing and genetic study) but patient refused and didn't came for further check up.

Discussion

Craniosynostosis is the premature fusion of one or more cranial sutures. Head shape anomalies or a syndromic diagnosis usually alerts the parent or pediatrician early in infancy to the potential underlying bony pathology.¹⁰ Ophthalmologic findings in cases of elevated ICP can be absent or difficult to detect. The initial ophthalmologic workup included a fundoscopic exam as well as an optical coherence tomography scan, which quantifies retinal nerve fiber layer (RNFL) thickness. This latter metric has been used as a noninvasive surrogate for conventional intracranial pressure measurements in children with pseudotumor cerebri; RNFL thickness was increased by nearly 75% on average when compared to measurements from healthy control subject.¹¹

Remodeling the cranial vault in an attempt to increase the intracranial volume and thus control intracranial hypertension. A case of craniosynostosis in whom cranial vault expansion was followed by the development of hind-brain herniation and

hydrocephalus. Craniosynostosis may fail to address the underlying aetiology of intracranial hypertension. Further more, both hydrocephalus and hind-brain herniation may develop following such surgery. Neither the increase in intracranial volume afforded by cranial vault expansion nor the shunting of hydrocephalus precludes the persistence of abnormal ICP.¹² With true syn-ostosis being differentiated from positional deformities and other normal variants. All patients with confirmed synostosis should be followed for evidence of progressive deformity, intracranial hypertension, and neuro-developmental problems.¹³

Conclusion

A 30 Years old primi gravida female came to opd for routine antenatal check up. As per usg finding she diagnosed as a lemon shaped skull and hydrocephalus.usg report convey to patient and advised for further evaluation but lost follow up of patient.

Most of the anomalies can be diagnosed antenatally if we do the anomalies scan around 18 to 20 weeks of gestation. So we should advice patient to come for regular antenatal visits and anomalies scan around 18 to 20 weeks of gestation. Once there is one anomaly try to search for another one. We should give folic acid 5 mg daily pre conceptionally for at least 3months to prevent neural tube defect.¹⁴

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