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Clinical Profile of Patients with Craniosynostosis: A Descriptive Study

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Craniosynostosis, Delayed Milestones, Profile.

A B S T R A C T

Craniosynostosis is important to recognise and treat because it can be associated with many complications affecting sensory, respiratory and neurological function. Our study was based on clinical evaluation and management of the patient, which was evaluated on Outpatient Department basis and called for surgery electively, except for the patients with raised intracranial tension and for the patients who had presented with acute visual loss. The majority of the patients had present with abnormal head shape (95.876%), followed by Proptosis (48.453%), Delayed Milestones (36.082%), Raised ICP (17.525%), Vision loss (17.525%), Fits (13.402%) and Upper Respiratory Tract Infection (3.092%). In one case of Coronal Synostosis, where the hearing was affected.

Introduction

Craniosynostosis, defined as the premature fusion of the cranial sutures, presents many challenges in classification and treatment. At least 20% of cases are caused by specific single gene mutations or chromosome abnormalities.

During infancy and childhood, the skull vault (calvaria) expands to accommodate the growing brain. This growth occurs predominantly at the narrow seams of undifferentiated mesenchyme, termed cranial sutures, which lie between different bones. The paired frontal and parietal bones are separated in the midline by the metopic and sagittal sutures, respectively; the frontal and parietal bones are separated by coronal sutures; and the parietal bones are separated from the single occipital bone by lambdoid sutures. Craniosynostosis describes the premature fusion of one or more of the cranial sutures: secondary distortion of skull shape occurs because of a combination of lack of growth perpendicular to the fused suture, and compensatory overgrowth at the non-fused sutures. The overall prevalence of craniosynostosis has been estimated at between 1 in 2100 and 1 in 2500 births (Lajeunie *et al.*, 1995). Craniosynostosis is important to recognize and treat because it can be associated with many complications

affecting sensory, respiratory and neurological function (Boulet *et al.*, 2008).

The precise aetiology of the craniosynostosis is not know, however various theories are put forth are studied under the following headings. The aetiology and pathogenetic heterogeneity; genetic evidence; the theories of Virchow (Virchow *et al.*, 1851); Moss (Moss, 1959) and park and powers (Park and powers, 1920); Metabolic disorders; drug teratogenicity: foetal head constraint; malformations, miscellaneous disorders with occasional synostosis, doubtful craniosynostotic association and experimental synostosis in animals.

Methodology

We have studied 97 cases of craniosynostosis. Our study was based on clinical evaluation and management of the patient, which was evaluated on Outpatient Department basis and called for surgery electively, except for the patients with raised intracranial tension and for the patients who had presented with acute visual loss.

Each casewere studied under following headings:

(1) History, (2)Clinical Assessment and (3) Investigations.

History emphasized on consanguinity and family history of similar deformity, along with social, physical and mental state of the patient. Most of the patients presented with abnormal head shape, delayed milestones, eye changes and with seizures.

Table.1 Deformities

Deformities	No. of Cases	Percentage (%)
Syndromal:		
Apert's	05	5.154
Crouzon's	08	8.247
Pfeiffer's	01	1.030
Total	14	14.431
Non-Syndromal:		
Unilateral Coronal (Right)	21	21.649
Unilateral Coronal (Left)	01	1.030
Brachycephaly (Coronal)	09	9.278
Dolicocephaly (Sagittal)	03	3.092
Trigonocephaly (Metopic)	12	12.371
Unilateral lamboid	01	1.030
Pansuture	28	28.865
Coronal + Sagittal	01	1.030
Coronal + Metopic	01	1.030
Clover Leaf	03	3.092
Microcephaly	01	1.030
Shunt for Hydrocephalus	02	2.061
Total	83	85.58

The above table shows the percentage of syndromal and non-syndromalsynostosis.

Table.2 Age and Sex distribution

Disease	No. of Cases	0-1 Months	2-6 Months	7-12 Months	13-24 Months	25-60 Months	61 Months	Male	Female
Coronal	9	-	3	3	-	3	-	6	3
Satittal	3	-	1	-	1	1	-	2	1
Metopic	12	2	4	1	2	1	2	11	1
Plagio (R)	21	1	3	6	5	5	1	7	14
Plagio (L)	1	-	-	-	-	1	-	1	-
Lambdoid	1	1	-	-	-	-	-	1	-
C + M	1	-	-	1	-	-	-	1	-
C + S	1	-	-	1	-	-	-	1	-
Microcephaly	1	-	1	-	-	-	-	1	-
Pansuture	28	-	1	2	3	5	17	13	15
Clover Leaf	3	2	-	-	1	-	-	1	2
Apert's	5	-	4	1	-	-	-	3	2
Crouzon's	8	-	-	3	-	3	2	5	3
Pfeiffer's	1	-	-	-	1	-	-	1	-
Secondary Synostosis	2	1	-	-	-	-	1	2	-
Total	97	7	17	18	13	19	23	56	41

The onset of the disease is within 6 months of life. However, the presentation of our OPD was under 5 years and 5 years onwards. Probably due to lack of knowledge of disease and its management, also due to delayed referrals.
In our study 56 (57.73%) were males and 41 (42.268%) were females. Youngest patient was 6 days old and eldest was 18 years old.
Ratio being Male : Female = 1.365 : 1.

Table.3 Age and Sex distribution in percentage

Disease	No. of Cases	0-1 Months	2-6 Months	7-12 Months	13-24 Months	25-60 Months	61 Months	Male	Female
Coronal	9	-	33.333	33.333	-	33.333	-	66.666	33.333
Satittal	3	-	33.333	-	33.333	33.333	-	66.666	33.333
Metopic	12	16.666	33.333	8.333	16.666	8.333	16.666	91.666	8.333
Plagiocephaly (R)	21	4.761	14.285	28.571	23.807	23.809	4.761	33.333	66.666
Plagiocephaly (L)	1	100	-	-	-	-	-	100	-
Lambdoid	1	100	-	-	-	-	-	100	-
Coronal + Metopic	1	-	-	100	-	-	-	100	-
Coronal + Sagittal	1	-	-	100	-	-	-	100	-
Microcephaly	1	-	100	-	-	-	-	100	-
Pansuture	28	-	3.57	7.42	10.714	17.857	60.714	46.428	53.571
Clover Leaf	3	16.666	-	-	33.333	-	-	33.333	66.666
Apert's	5	-	80	20	-	-	-	60	40
Crouzon's	8	-	-	37.5	-	37.5	25	62.5	37.5
Pfeiffer's	1	-	-	-	100	-	-	100	-
Secondary Synostosis	2	50	-	-	-	-	50	100	-

Males predominance is seen in synodromes, coronal, metopic and pansuture, secondary synostosis, however in (R) Plagiocephaly females outnumbered males.
The following table shows family history and consanguinity, in number of cases and percentage.

Table.4 Disease

Disease	Family history	Consanguinity
Coronal	1 (11.111%)	1 (11.111%)
Sagittal	-	-
Metopic	2 (16.666%)	-
Pansuture	3 (10.714%)	1 (11.111%)
Clover Leaf	-	-
Apert's	-	1 (20.000%)
Crouzon's	1 (12.500%)	1 (11.111%)
Pfeiffers	-	-
Total	7	4

Family history was positive in 1 (11.111%) case of Coronal Synostosis (maternal and paternal uncles had similar disease). In Metopic Synostosis 2 patients (16.666%) had similar disease in their cousines, of 28 PansutureSynostosis, 3 cases had similar disorder in their family (1 father, 1 sister and 1 brother).

History of Consanguinity was noted in 4 cases one each in Coronal, Pansuture, Aperts and Crouzons disease. We had two pairs of twins. In first pair – one was normal and other suffered from Plagiocephaly. Second pair of twins – were born prematurely at 7 ½ months, first died after 6 days from birth asphyxia and neonatal jaundice, second one presented with pansuturesynostosis.

Table.5 Symptoms

Disease	Abnormal Head	Delayed Milestone	Proptosis	Raised ICP	FITS	Vision Loss	URTI
Coronal	9	5	3	1	2	-	1
Sagittal	1	1	1	1	-	-	-
Metopic	12	6	2	1	1	-	-
Plagiocephaly (R)	21	7	8	1	2	1	2
Plagiocephaly (L)	1	-	-	-	-	-	-
Lambdoid	1	-	-	-	-	-	-
Coronal + Metopic	1	1	-	-	-	-	-
Coronal + Sagittal	1	1	1	-	1	-	-
Microcephaly	1	-	-	-	-	-	-
Pansuture	28	9	21	10	5	13	-
Clover Leaf	3	-	3	-	-	-	-
Apert's	4	1	3	1	-	-	-
Crouzon's	7	2	5	2	1	2	-
Pfeiffer's	1	-	-	-	-	-	-
Secondary Synostosis	2	2	-	-	1	1	-
Total	93	35	47	17	13	17	3

URTI = Upper Respiratory Tract Infection.

The majority of the patients had present with abnormal head shape (95.876%), followed by Proptosis (48.453%), Delayed Milestones (36.082%), Raised ICP (17.525%), Vision loss (17.525%), Fits (13.402%) and Upper Respiratory Tract Infection (3.092%). In one case of Coronal Synostosis, where the hearing was affected.

Examination of the patients for head shape, suture state, eye changes, deformity of face and other associated anomaly, X-ray skull and CT scan was done in almost all cases. Postoperatively skull x-rays were repeated after a week, every 3 months for first year and at yearly interval for comparison and for records. The cases were divided into syndromal and non-syndromal.

Results and Discussion

The precise aetoilogy of craniosynostosis is not known. Most often is sporadic, but predominantly autosomal dominant and sometimes recessive. There are recognized syndromes associated with mendelian inheritance: (1) like Crouzon's and Apert's syndrome (dominant) while carpenter syndrome is clearly an autosomal recessive trait, (2) mutation is also been implicated in different generations. In our study, mother and child having Crouzon's disease, father and child having pansuturesynostosis, there was a similar problem in paternal uncle and grandfather. We had two cases of trigonocephaly in maternal cousins and coronal synostosis in maternal and paternal uncles. In all these cases genetic studies were in conclusive.

The incidence reported from different countries varies from as low as 0.4/1000 to high as 10/1000. There is overall male preponderance according to the literature. In our study 56 were males and 41 were females. The frequency of twinning in several surveys of craniosynostosis. Percentage varies from a high at 19.4% brestlen's series, to a low of 2.4% in David *et al.*, series (David *et al.*, 1986).

In our series, we had two pairs of twins which amounts to 2.061% However there is no mentioned of consanguinity in the literature. We had 2 cases giving history of consanguineous marriage. Following table showing the number of cases in comparision with Montaut and Stricker series (1997) (Montaut Sticker Les, 1977).

Disease	Montaut & stricker series	Our series
Coronal	34 (29.310%)	09 (9278%)
Sagittal	35 (30.172%)	03 (3.092%)
Metopic	13 (11.206%)	12 (12.371%)
Plagiocephaly (R)	---	21 (21.649%)
Plagiocephaly (L)	---	01 (1.030%)
Lambdoid	13 (11.206%)	01 (1.030%)
Coronal + Metopic	---	01 (1.030%)
Coronal + Sagittal	---	01 (1.030%)
Microcephaly	---	01 (1.030%)
Pansuture	32 (27.586%)	28 (28.865%)
Clover Leaf	---	03 (3.092%)
Apert's	08 (6.896%)	05 (5.154%)
Crouzon's	21 (18.103%)	08 (8.247%)
Pfeiffer's	---	01 (1.030%)

Conclusion

Family history was positive in 3 cases of PansutureSynostosis (10.714%), 2 cases of Metopic Synostosis (16.666%), 1 each in Coronal and Crouzon's syndrome. However, Consanguinity was noted in 4 cases, Consanguinity is not described in the literature.

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