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DIAGNOSING CRANIOSYNOSTOSIS – "THE EARLIER THE BETTER" – A CASE STUDY

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ABSTRACT

Craniosynostosis is a developmental craniofacial anomaly, resulting in impairment of brain development and abnormally shaped skull. The main cause of craniosynostosis is premature closure of one or more cranial sutures. Delayed diagnosis be associated with abnormalities in brain development including raised intracranial pressure and neurocognitive development impaiments. A descriptive phenomology approach was used to collect data from parents of children, children who was diagnosed with craniosynostosis after the age of 10 years and above by the physician were included in the study. The collected verbatim was transcribed into meaningful inference. Data was collected till data saturation from series of four patients. The major reason identified were Majority with Lack of knowledge about the disease, then a small majority expressed financial constrains for treatment, few were anxious about the outcome, a few reported lack of awareness among health care professional regarding the disease, Societal stigma of the disease.

KEYWORDS: Craniosynostosis, Crouzons syndrome, Aperts syndrome, development, diagnosis, treatment.

INTRODUCTION

Congenital disorders can be defined as structural or functional anomalies that occur during intrauterine life. Also called birth defects, congenital anomalies or congenital malformations, these conditions develop prenatally and may be identified before or at birth, or later in life. An estimated 6% of babies worldwide are born with a congenital disorder, resulting in hundreds of associated deaths.[1] Craniosynostosis is one of the rare developmental anomaly which occur as a consequence of abnormal and non- physiological sutural fusion.[2]

Craniosynostosis is a congenital craniofacial disorder with a prevalence of 3.1-6.4 per 10,000 live births worldwide.[3] In a newborn the membranous bones of the cranial vault are separated by the intervening sutures. Such arrangement enables the infant's skull to pass more easily through the birth canal and allows the compensatory growth of skull during brain growth. When one or more sutures are prematurely closed, the compensatory growth starts perpendicular to the parent sutures since the brain still grows and expands in the direction of lower resistance. The result is an abnormally shaped skull and also, in more severe cases, increased intracranial pressure (ICP), as well as sensory, respiratory and neurological dysfunctions.[1] The diagnosis of a typical craniosynostosis is usually clinical and it is commonly diagnosed in the first year of life. It usually occurs as an isolated condition, but may also be associated with other malformations as part of complex syndromes. Although the exact cause remains unclear, genetic factors are thought to play an important role. Mutations in genes such as FGFR2 and FGFR3, which disrupt the normal development of the skull, are suspected. Environmental factors and various insults during pregnancy can also contribute to the occurrence of the disease. An accurate diagnosis is crucial for treatment. Imaging studies such as ultrasound, computed tomography, magnetic resonance imaging, and three-dimensional reconstructions play a crucial role in visualising the prematurely fused sagittal suture.

The clinical assessment determines the the following: whether a craniosynostosis is present, whether there are additional features suggesting an associated syndrome, and whether urgent or elective management is required.[2]

Recently, CDC reported on important findings from research studies about some factors that increase the chance of having a baby with craniosynostosis: Maternal thyroid disease — Women with thyroid disease or who are treated for thyroid disease while they are pregnant have a higher chance of having an infant with craniosynostosis, compared to women who don't have thyroid disease.[2] Certain medications — Women who report using clomiphene citrate a fertility medication just before or early in pregnancy are more likely to have a baby with craniosynostosis, compared to women who didn't take this medicine.[3] And the other risk factors like genetic mutation smoking alcohol and certain medication contribute to craniosynostosis.

When untreated it results mainly visible feature of abnormal shape of the head and dysmorphic facial features including anomalies affecting the eye. Along with the cosmetic effect the child will be ending with a physiological defects like developmental delay, respiratory and neurological dysfunction, facial abnormality, sensory dysfunction, anomalies affecting the eye, and psychological disturbances,. Some of the factors that can lead to delay in diagnosis and treatment are healthcare unavailability, knowledge of the pediatrician and healthcare workers, financial instability, cultural beliefs and parental fear and anxiety on the treatment strategies including surgery. Thus, early diagnosis, expert surgical techniques, postoperative care, and adequate follow-up are of vital importance in treating craniosynostosis.

Method

A descriptive phenomology approach was used to collect data from parents of children, children who was diagnosed with craniosynostosis after the age of 10 years and above by the physician were included in the study. A verbal Informed consent was obtained from the parents and assent from the child before collecting data. A interview schedule was using a semi structured questionnaire was used to collect the data from the parents and the child. The collected verbatim was transcribed into meaningful inference. Data was collected till data saturation from series of four patients.

RESULT

Case presentation

Case 1

Presenting the case of 15year old child who got diagnosed in the age of one and half year. Certain risk factors were present at the time of pregnancy like maternal stress, as the mother

was alone at home during the antenatal period and the workload was more. First child was a preschooler and the blood group of mother, father and the child was O+ve.

Antenatal and Intranatal period was uneventful. It was a prolonged delivery followed by vaccum extraction. On delivery amniotic fluid aspiration was present. Breastfeeding was initiated. After delivery exophthalmia was elicited. Physicians have recognised the abnormality and neonatologist told that there is a possibility of chromosomal abnormality.

Even the parents asked that antenatal, intranatal and postnatal events were uneventful and then how this could be missed.

At birth abnormality was detected on physical examination by the physician and parents especially exophthalmia. The neonatologist told there is a possibility of chromosomal abnormality. They by themselves went to ophthalmic consultation at Madurai. The physician told that there was an abnormal proportion of eye ball and eye socket for the baby and they referred the baby for tertiary care hospital.

Since the private tertiary care expenses are not affordable, they went to Government medical college and after investigation diagnosed as Crouzons syndrome.

Also parents noted that as the baby aged the eye of the baby was not properly closing especially during sleep. There surgical management of Intraoptic Rim Augmentation for the eye was done. After surgery there was a decrease in the size of eye ball and prominent developmental delay was present for the child. The child had developmental delay of six months and it was observed that there is disproportion between the head and the body.

Small mass was found in the forehead and they consulted in the same Government medical College where the previous surgery was done. The physician told it may be the post surgery edema and symptomatic management was done. The edema didn't subside. And they noted that the nasal opening was small, teeth eruption showed abnormal malposition with huge gap, mouth breathing so they consulted the physician. They insisted and referred to private tertiary care hospital by two years.

On examination and diagnostic investigation in the hospital they found fragments of bone was still present and pharmacological management was done and the patient was asked to

review for the maxillofacial surgery department. At eight years no surgery was done. By the age of twelve years surgical management was done under maxillofacial surgery department.

At fifteen years, the patient tested intelligence quotient level and twenty-five percent disability was identified. He is an average learner. He is interested in sports.

At present vitiligo is present for the child and is having concerns regarding this. While communicating with the patient slurred speech is present.

On conclusion we asked them what are the factors that you think resulted in the delay in diagnosis and treatment of your child. And they replied that mainly the financial instability in the family and lack of awareness among the health professionals.

Case 2

A twenty three year old female child from who is a known case of Crouzons syndrome got diagnosed in the age of nineteen. The risk factors for this disease is mainly blood group of mother, father and child is B+ve and the maternal stress due to the work as she was a unskilled field worker.

During antenatal period immunization was done as per government protocol and supplementation was done regularly. No antenatal scanning was done during the antenatal period. It was a traditional home dias assisted delivery. Postnatal events were uneventful. Breastfeeding started and the immunization was done regular. Routine checkup of the child was done at the Government primary health centre.

By the age of five years, they noticed abnormalities in the face of the child and the physicians and neighbours told them it is normal and will subside slowly.

In seventeen year of age, the eye seems abnormal to her brother and took advice from his friend who was pursuing masters in medicine and he suspected some kind of syndrome for the child. Thereafter they went to a local hospital in North India and undergone X ray and they diagnosed it as for a possibility of Treacher Collins Syndrome. No further management was done.

Later they went to private tertiary care hospital at nearby city and undergone clinical examination and radiological examination. Then diagnosed as Crouzons syndrome. But the

physician has not explained about the syndrome much. He had only advised to perform surgery.

The family was advised to visit the tertiary care hospital in South India but due to the long distance and language barrier they preferred to have consultation at a hospital in nearby city.

There they consulted the neurosurgery department and they advised to visit the tertiary care hospital in South India as they have an expert team for Craniomaxillary facial surgery and experience in treating the disease.

By the end of eighteen years of age they visited the Cranio-Maxillo Facial surgery department at the tertiary care hospital in south India and started with the management and treatment.

On examination they identified that the patient had pseudoproptosis, supra orbital ridge is retrodisplaced, severe maxillary hypoplasia, mandibular retrognatism, anterior crossbite and she underwent CT scan which showed malshaped cranial vault, small posterior fossa with foramen magnum compression and had undergone posterior calvarial distraction.

Later she had developed headache, nausea and vomiting and got admitted following distraction. The headache was suddenly in onset and moderate in intensity. After the surgery by around nineteenth year as a part of the second stage of treatment she had undergone tarsorraphy and lefort 3 advancement after examination which showed flattened forehead, antimongoloid slant proptosis, orbital dystopia and midface deficiency.

The patient had undergone bilateral sagittal split osteotomy advancement genioplasty, when she was presented with elongated face, increased lower facial height, incompetent lips, interlabial gap. Also she developed complaints of incomplete closure of eyes during sleep and had underwent Fronto orbital advancement+ malar augmentation.

She is a below average student with an intelligence quotient of sixty to seventy percent. From the brothers view she is having an insecurity about her physical features and had faced bullying at the school as a result she has a few friends. She sometimes feels shameful and will not be attending any functions.

On conclusion we asked them what are the factors that you think resulted in the delay in diagnosis and treatment of your child. And they replied that mainly the financial instability in the family and lack of awareness among the family members and health professionals, lack of adequate support system.

Case 3

A case presented in the neurosurgery OPD aged eighteen years diagnosed at the with Crouzons syndrome at 14 years of age. Risk factors considered are financial issues, family issues among the relatives and blood group of mother, father and child O+ve.

Antenatal, intranatal and postnatal events are normal. Mother had stress from the family and financial issues during the antenatal period. At birth exophthalmia was present and routine investigation was done regularly for the child.

Due to exophthalmia an ophthalmic surgery was done at nine months of age in a private tertiary care hospital. Child was presented with growth retardation where height and weight are not appropriate for the age.

When he was sixteen years old dental consultation was done in a government medical college and got referred to private tertiary care hospital. By seventeenth years of age they took consultation in a tertiary care hospital but surgery and treatment was postponed due to financial issues. Later treatment and surgery was initiated.

In the year seventeenth year, patient was brought to Craniomaxillary facial surgery with complaints of inability to close eyes and experiencing breathing difficulty. Clinical and radiological examination was done and diagnosed with Crouzons syndrome.

On clinical examination marked midface and frontal hypoplasia, severe proptosis with subluxation of eye ball, crowding in maxillary arch, hearing difficulty present and underwent monobloc distraction of frontomaxillary region in the same year.

From his mothers view he is an average student in the class and had proper development. After surgery he is alright and doing better in day to day activities.

Case 4

Seventeen year old male child came to neurosurgery OPD with Aperts syndrome. Risk factors that caused this condition are blood group of mother, father and child is B+ve and stress during pregnancy period. Maternal stress of work as she is a contract based worker in a construction company and worked till seven months of pregnancy.

Antenatal, intranatal and postnatal events are uneventful. At birth he had clubbing of the fingers in upper and lower limb and exophthalmia.

When he was 9 month of age, he underwent syndactyly release as he was having clubbing of fingers and diagnosed with bilateral upper and lower limb syndactyly. And was doing good.

In nine years of age, patient was brought to OPD with complaints of midface placed backward and difficulty in articulating words. On examination he was having exophthalmia, hypertelorism, orbital dystopia, off set right ears, incompetent lips, root of nose deviated to left, midface retrusion, class 3 malocclusion, narrow deep palate. And thereafter he underwent Fronto orbital advancement+ bipartition surgery and hypertelorism correction. In the mid of nine years of age the rigid external device was removed. And by end he underwent palatoplasty as he was having cleft of secondary palate, bifid uvula, speech and articulation problem, mild hypernasality.

In ten years of age again consulted in OPD in with complaints of both hands 2-5 fused digits on either side. And left first web space deepening on hand + first web space creation on toes bilaterally and skin grafting of first web space of bilateral feet under general anaesthesia done. He had history of web space creation at outside hospital. Syndactyly of all toes present and left foot toes 3,4,5 upturned.

In ten years of age, patient presented with bilateral temporal hollowing, crooked nose, down slanting lateral canthus bilaterally, irregularities in frontal region, high arched palate. And underwent revision cranioplasty and rhinoplasty.

In the fifteen year of age, on clinical examination it was observed that patient had asymmetrical antimongoloid slant, broad and short nose, incompetent lips, constricted maxillary, crowded dentition, hypertrophic palatal mucosa, palatally placed soft tissue and underwent hypertrophic palatal tissue debulking and extraction of teeth.

DISCUSSION

This study was conducted to identify the knowledge of parents in the early diagnosis and treatment of craniosynostosis. Both the cases showed that the defect present in the pregnancy period was not at the earliest. The case clearly shows that there was a delay in early diagnosis and treatment and how the delay had lead to the defects including in the brain development of the child even though much remarkable changes in the level of intelligence but may be if surgery was performed early their academic performance would have been better. The cases also indicate the need of further education about craniosynostosis for health visitors to promote early detection.

The important remarkable risk factors observed was all parents and children had same blood groups.

The most frequent challenges faced in finding information were due to not being aware of the reliable sources of information and was not aware of identifying defects earlier at the time of pregnancy itself. This shows the need for healthcare providers to educate and identify the defects in the earlier stages to prevent the defects.

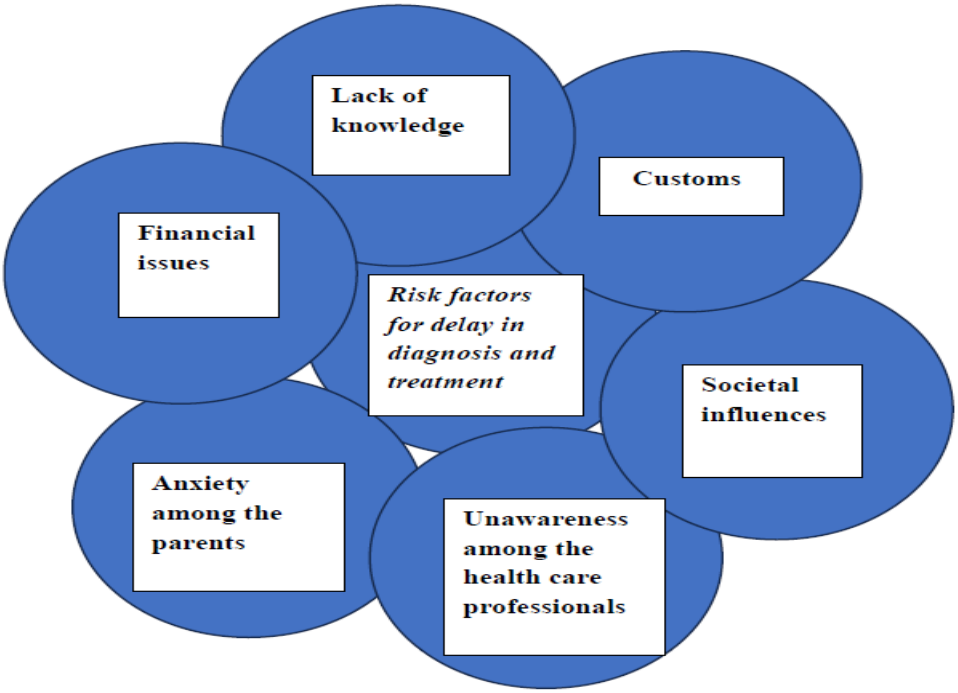

Fig. 1: Major reason identified for delay in diagnosis of patients with craniosynostosis.

The major reason identified were Majority with Lack of knowledge about the disease, then a small majority expressed financial constrains for treatment, few were anxious about the

outcome, a few reported lack of awareness among health care professional regarding the disease, Societal stigma of the disease.

CONCLUSION

Early diagnosis is better for good growth and development of children overcoming the above mentioned factors will aid in early diagnosis which will have the best outcome.

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