Rare disease

Crouzon syndrome: a social stigma

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Summary

Crouzon syndrome is a rare genetic disorder caused due to genetic mutations. It is characterised by partial hearing loss, dry eyes, strabismus and underdevelopment of the upper jaw with facial deformities and malocclusion. These facial deformities greatly affect the social and emotional development of the affected child. The present case report highlights the social problems faced by a child suffering with Crouzon syndrome.

BACKGROUND

Crouzon syndrome is a rare genetic disorder estimated to occur in 1.6/100 000 population. It is caused due to genetic mutations in the fibroblast growth factor receptor (FGFR) genes—FGFR2 genes on chromosome 10 and FGFR3 on chromosome 4.^{1 2}

Crouzon syndrome varies in severity in affected individuals. The affected individuals might exhibit partial hearing loss, dry eyes, strabismus and underdevelopment of the upper jaw which produces facial deformity and malocclusion.^{3 4} The malformation of the face creates social stigma which affects their emotional development. The present article attempts to highlight the social problems faced by the children afflicted with this disorder.

CASE PRESENTATION

An 11-year-old female child patient reported to our outpatient department with the complaint of inability to eat due to pain in the lower left side of the mouth. Extraoral examination revealed the presence of strabismus and underdeveloped midface (figure 1). Exophthalmos was

obvious on lateral facial profile view (figure 2). On intraoral examination, left lower primary second molar (75) was found to be carious and advised for extraction. Further medical history given by her parents revealed that she was not having her meals properly, which was initially supposed to be because of the aforementioned tooth. After the extraction, the patient was recalled after 3 weeks for a routine check-up.

On the subsequent appointment, the parents still narrated her not having meals properly, lack of interest in anything and dissent in going to school daily. Her parents reported that she would sit in a dark corner of the room and would not talk or eat at all for 2 days at a time.

After recording the complete medical, dental and social history, the child was scheduled for a general conversation session without the parents. Initially, the child was reluctant and very scared but on the third appointment the child slowly became more outspoken, less scared and started narrating to the clinician.

Figure 1 Patient's photograph showing strabismus and underdeveloped midface.

Figure 2 Lateral facial profile exhibiting exophthalmos.

It was noticed that the child faced a lot of problems in adapting herself to the social environment. She was treated as an outcast not only by the children of her age but also by her teachers. Everyday she would be subjected to very rude remarks. Since she had exophthalmoses and proptosis, her eyes bulged out making her look different; some of the kids would try poking her in the eye with sharp objects. She would be ridiculed in the streets and at home by her older siblings too. No one around her would like her company. She was scared to go out of her home anymore and would want to hide in the dark corners hiding her face.

INVESTIGATIONS

CT scan of the patient was performed. The radiological evaluation of the CT scan stated craniosynostosis with brachycephaly and scalloping of the inner table of the calvarial bones with hypertelorism, proptosis and hypoplastic visualised facial region and dilacerated lateral ventricles (figures 3–5). The clinical findings were also suggestive of Crouzon syndrome as evidenced in CT scan.

TREATMENT

The child was referred to a psychologist and the parents were also counselled with the child. They were advised to understand the problems their child faced and that the child needed a lot of emotional support and sympathy from the family, so as to build up her self-esteem and confidence to lead a good life ahead.

OUTCOME AND FOLLOW-UP

After months of psychological counselling, a lot of improvement was seen. The child slowly started adjusting herself to the social environment. She was shifted to the school for disabled children where she came to know about other children who were also special in their own ways. Her interest in resuming studies slowly built up too. Her diet gradually improved.

Figure 3 CT scan showing brachycephaly with scalloping of calvarial bones.

Figure 4 CT scan showing hypertelorism with proptosis and underdeveloped midface.

DISCUSSION

Child psychology is an integral part of paediatric dentistry which is often overlooked and taken for granted. It is thought that the children are too immature and might probably forget what happens in childhood and hence childhood experiences are of no consequence whatever in playing a role in their future development. A number of researches have shown that the prepubertal and adolescent period in a child's life is one of the most important phases of life where in the future mindset of the child's emotional development takes place, based on this crucial time period's events.^{5 6}

Figure 5 CT scan showing dilacerated lateral ventricles in the cerebrum.

The children with special needs are completely dependent emotionally and sometimes physically on their family members and friends to cope with the emotional stresses which they have to undergo in day-to-day life.^{6 7}

There is no definitive cure for Crouzon syndrome. With the continual familial support, physical rehabilitation can be done for the patient to some extent by the surgical advancement techniques,⁸ but still the support and the confidence instilled in the child would help him to acquire coping strategies to lead a near-normal life.⁹

Learning points

- ▶ Crouzon syndrome and others, not only affects the child's physical appearance but also have an adverse effect on the personality development of the affected child, making them vulnerable to emotional breakdown and disinclined to live.
- ▶ As there is no treatment for Crouzon syndrome, such special children should be treated with tender, love and care by the society as they need more help and support to lead a near-normal life than the otherwise normal children.
- ▶ Psychological counselling of such special children may be advantageous to increase their self-confidence and build their self-esteem, which indeed may help them to cope with the differences faced by them every so often by the community.
- ▶ As the parents of the affected children also face the emotional disturbance and social embarrassment, their counselling may be helpful to make them understand the sufferings of their child and not to impose their frustration in the form of child abuse and neglect.

Competing interests None.

Patient consent Obtained.

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