International Journal of Practical Nursing
Volume 8 Number 1, January - April 2020
DOI: http://dx.doi.org/10.21088/ijpn.2347.7083.8120.6

Case Report

Rare Genetic Dissorder: Crouzon Syndrome

Gyanderi Tomar[1], Vasantha Kalyani[2]

Author Affiliation: [1]Nursing Tutor, [2]Associate Professor, College of Nursing, All India Institute of Medical Science, Rishikesh, Uttarakhand 249203, India.

How to cite this article:
Gyanderi Tomar, Vasantha Kalyani. Rare Genetic Dissorder: Crouzon Syndrome. Int J Practical Nurs. 2019;8(1):33–39.

Abstract

Crouzon syndrome is a rare-case found in children resulted from genetic disorder. It is characterized by premature closure of cranial sutures, most commonly coronal and sagittal, that is the most common cause of skull abnormality. Common characteristics are vision problem resulted from exophthalmos, beaked nose and underdeveloped upper jaw. In worldwide it occurs in approximately 1 in 25000 births.[1] It is the most common syndrome of craniosynostosis group. Crouzon syndrome is caused bymutationin the extracellular immunoglobulin III domain of the Fibroblast Growth Receptors 2 (FGFR2) gene. In this type of syndrome there is no hand and/or foot abnormality that makes it different from other craniosyntosis syndrome.[2] The only effective treatment is multiple staged surgeries. Here presented a case study on a 2 year old boy with Crouzon syndrome.

Keyword: Suture; Craniosynostosis; Brachicephaly; Crouzon syndrome; Mutation.

Introduction

Baby X, 1 year 9 monthold male child diagnosed as craniosynostosis and brachycephalic due to Crouzon syndrome. That is a genetic disorder resulted premature fusion of skull bone. On 18th April 2019 parents of Baby X came to AIIMS Burn and plastic OPD. On 2nd July 2019 at 2:45 pm baby X was admitted in burn and plastic surgery ward. A collaborative management was planned to treat him. On 13th July 2019 fronto-orbital advancement surgery and cranioplasty was done under general anesthesia and patient intubated with ET tube no 4 mm. On 4th August ventriculoperitoneal shunting was done to treat Hydrocephalus. On 1st September 2019 concerned doctor planed for permanent lateral tarsorraphy.

Treatment started with Inj Meropenam 300 mg (TDS), Inj Colistin 2 lac (TDS), Inj Eptoin 25 mg (BD), Inj levipil 130 mg (BD), Tab Diamox 30 mg (BD), Tab Clobazem 5 mg (OD).

On 14 September 2019 discharge was planned. At that time general condition and vitals were stable and hygiene maintained. No any other fresh complain noted. Health educations given to the parents of patient and advised for Follow-up.

Fig. 1:

Corresponding Author: Vasantha Kalyani. Associate Professor, College of Nursing, All India Institute of Medical Science, Rishikesh, Uttarakhand 249203, India.

E-mail: vasantharaj2003@gmail.com

Received on 26.11.2019, **Accepted on** 19.03.2020

History Collection

There was no family history of congenital abnormality. There was no consanguineous marriage between the parents of baby X. The child was second in birth order. His elder brother was 7 year old and he was normal. Delivery of baby X was at full tern and it was normal delivery but the baby was with bulgy eyes. At birth he was 3.7 kg. After 2–3 month of birth parents took him to the hospital for the treatment of protruded eyes and from therethey referred him to eye specialist. Then again he was referred to one of the super specialty hospital. There eye test was done and referred him to neurosurgeon and diagnosed as craniosynostosis and Doctor suggested for surgery.

Family Pedigree: 4 Generation

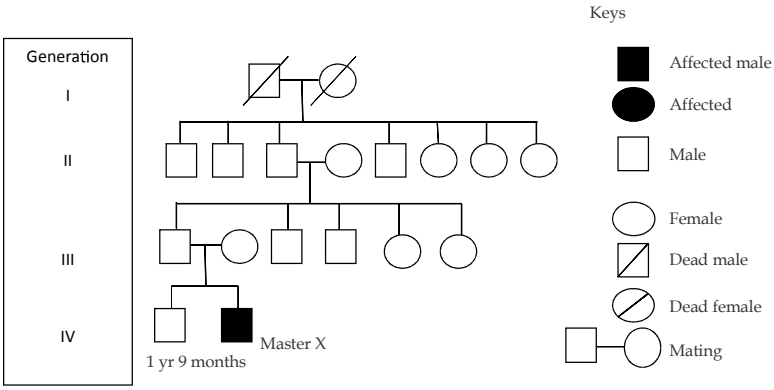


Fig. 2: Family Pedigree: 4 Generation

Physical Examination

General appearance: normal body built and Conscious.

- Skin: Intact without any spot and rashes.
- Head: Enlargement of head and prominent forehead
- Face: Depressed mid facial region.
- Nose: Nasal bridge depressed and nose tip is upward
- Eyes: Bulgy eyes and no alignment
- Neck: Normal length
- Chest: Transverse diameter was greater than anterioposterior diameter
- Abdomen: Soft, palpable
- Extremity: All digits present, equal length and symmetry.
- Vital sign: Temperature 98°F

Pulse	115 beats/ min
Respiration	24/mint
BP	97/61 mm Hg
SPO_2	99%

Disease Condition

Table 1: Disease Condition

Topic	Book Picture	Patient Picture
Definition	The premature closure of the skull sutures, leading to interference with the proper brain growth, is termed craniosynostosis or, to be more exact, craniosynostosis.[3]	
Epidemiology	Birth prevalence: Craniosynostosis have an estimated prevalence of 1 in 2500 live birth worldwide in early studies and 1 in 60,000 in later studies and 16.5 per 1,000,000 live births in united states. Crouzon syndrome accounts approximately 4.8% of all cases of craniosynostosis. About 70-80% of cases are non-syndrome.[1,4]	
Classification		
	1. Oxycephaly: This mean fusion of coronal and in some cases all the sutures. The head may be antero-posteriorly flattened and elongated transversely and upwardly. This is called acrocephaly.[3]	Acrocephaly present

Topic	Book Picture	Patient Picture
	2. Scaphocephaly: Here the saggital suture is fused. As a result, skull grows antero-posteriorly and thus assumes an elongated appearance resembling a boat.[3]	
	3. Plagiocephaly: Asymmetrical fusion of sutures leading to asymmetrical skull.[3]	
Types		
	Syndrome: Autosomal dominant with complete penetrance and variable expressivity[5]	
	Nonsyndrome: Arising from fresh mutation[6]	No one is having any genetic disorder in family
	Crouzon syndrome is caused by. Mutation in FGFR2 (Found in 50% patient) or FGFR3 gene.[7]	
	With fresh mutation (Found in 50% cases).[7]	Genetic changes resulted from fresh mutation
Causes	Parent with crouzon syndrome having 50% chances to have a baby with crouzon syndrome[8]	
	Paternal age (there is association between older paternal age and Sporadic crouzon syndrome)[9]	
Pathophysiology	• Fibroblast growth factor receptor-2(FGFR2) gene mutation[10] • Premature synostosis of coronal, sagittal and rarelylambdoidal suture begins(1st year of life)[10] • it may complete in 2nd and 3rd year resulted deformity[10] • Increased intracranial pressure[10] • cloverleaf skull (Brain protrudes from open anterior and parietal fontanelles)[11] 1. **Craniosynostosis:** Premature closure of sutures of the skull results changes in the shape of the head and results increased pressure on the brain. This changes appearance of head tall and flat from the middle portion of the faces upward.[12] 2. **Midfacial hypoplasia:** Decreased growth of the middle of the face, causing a sunken facial appearance. This can also cause potential airway obstruction, sleep apnea and a concave facial profile.[12] 3. Which results[8,13] • Shallow orbits • Midfacial-hypoplasia • Foreshortened nasal dorsum • Exophthalmos • Maxillary hypoplasia • Occasional upper airway obstruction	
Clinical features		
	Head[14]	
	In early suture fusion head appears short and broad (Brachicephaly)	Head is transversely broad
	In other patient head may appear long and narrow (Scaphocephaly)	Forehead is prominent
	Prominent forehead	
	Eyes[15]	
	Exotropia	Eyes don't look in the same direction at the same time
	Dystropia	Eye are not in same horizontal line
	Exophthalmos	Both eyes are bulging out
	Approximately 46% of patient experience poor vision	Poor vision
	Optic nerve atrophy found in 22%	Blurred vision present

Topic	Book Picture	Patient Picture
	Blindness found in 7%	Not present
	Nose[14]	
	Curved nose	Bridge of nose is downward and tip upward
	Flat or underdeveloped mid facial regions	Flattened mid facial region
	Ear[14]	
	Conductive hearing deficit	
	Artesia	
	Oral cavity and dental anomalies[14]	
	Lateral palatal swelling (in half of the patient)	
	Cleft lip and cleft palate (rare)	
	Mandibular prognathism	Protrusion of lower jaw
	Hypoplastic maxilla	Underdeveloped maxillary bone
	Reduced dental arch width	Dental crowding is present
	Ectopic eruption of maxillary first molars	
	Malocclusion	Misalignment of teeth present
	Central nervous system[8,16]	
	Progressive hydrocephalus	Enlargement of head is present
	Headache	
	Vomiting	
	Seizures	Sudden jerky movements with clenching teeth
	Intracranial pressure alterations	
	Facial palsies	
	Sensory impairment	
	Ataxia	
	Spasticity	
	Abnormality of breathing, swallowing or sleep	Shallow breathing pattern
Investigation		
	X-ray[17]	Done to rule out proper position of shunt
	CT scan[17]	CT of head rule out premature fusion.
	Genetic testing (blood/saliva)[3]	
	MRI[18]	

Fig 1 (a) Fig 1 (b)

Fig. 1: X-ray chest and head to rule out position of shunt.

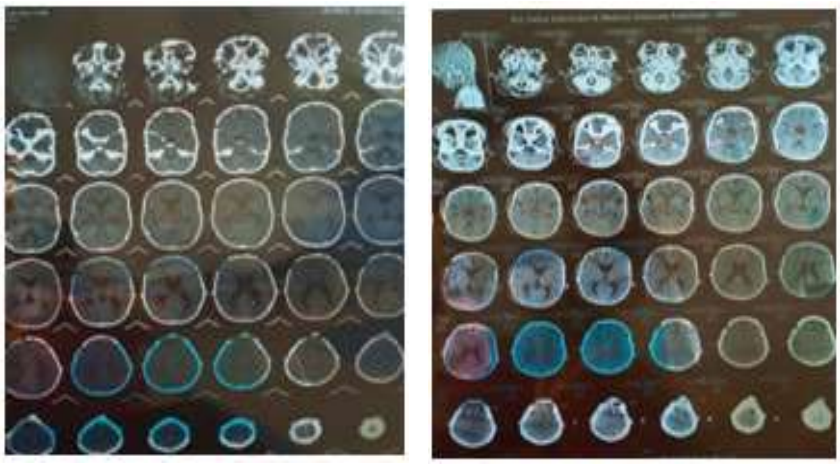


Fig. 2: CT Head to rule out premature

Management

Management of the patients who is suffering from Crouzon syndrome requires multidisciplinary approach of care that includes plastic surgeons, Neurosurgeons, Pediatricians, Otolaryngologists, Geneticists, and Ophthalmologists to work in collaboration.[17]

Craniofacial centers as they have great experience with Syndromic craniosynostosis. Early diagnosis and treatment help to prevent further problems, and the complications.

- Early detection and management of Eye problem:

  Early detection of eye problem: Early detection of changes in eye sight can be treated early. Refractory errors, strabismus etc are few common problem of eye in crouzon syndrome and these can be treated early.[42]

  Treatment of visual impairment: A high prevalence of visual impairment is reported in patients with Crouzon syndrome. Treatment of strabismus and refractive errors should be a priority for ophthalmologists.[41]

- Early detection and management of airway:

  Early detection of nose problem: It is important to find out airway obstruction due to changes in nasal bone and palate.

  Treatment of respiratory impairment: A nasal continuous positive airway pressure device may be needed to relieve airway obstruction.

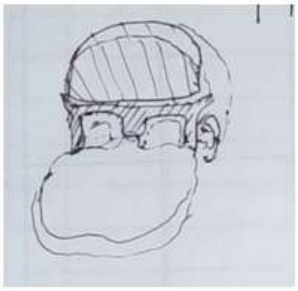
- Early detection and management of hearing loss:

  Early detection of ear and hearing problem: A patient with crouzon syndrome may have various pathological features of ear. Early otologic and audiologic assessment is indicated to detect obstructive, sensorineural and mixed hearing loss.

  Treatment of hearing impairment: Microphones, cochlear implant and Follow-up are some measures to treat hearing impairment.

- Management of speech by speech therapy.

Treatment:[19]

Craniotomy	Done by burr hole and giggly saw to repair skull bone
Endoscopic strip craniectomy	
Fronto-orbital advancement	Osteotomy cuts made to remove frontal bone. Orbital band was cut and brought it forward and fixed it SS wire. Flap closed with running Nylon suture (3-0).
	
Cranioplasty	Done to repair deformity

Nursing Management

Assessment: Obtaining complete history to assess for symptoms that indicates genetic disorder. A detail history of family that covers three generations has been taken. Physical examination of baby as well as parents been done and attention given to skull, eyes, nose, ear and hard palate. Mother's pregnancy history has been taken.

Nursing Diagnosis: Deficient knowledge related todeformity and treatment regimen.

Goal: To enhance knowledge regarding deformity and its treatment

Nursing Intervention:

- Support and teach the patient to adhere to treatment regimen
- Schedule regular follow-up appointment
- Teach cause of disease, its process and consequences.
- Counsel the patient fordisease its cause impact on life style, and focus on modification of lifestyle.

Nursing diagnosis: Disturbed head and face image Related to Premature fusion of skull sutures

Goal: Discuss with the family to generate the acceptance of situation

Intervention

1. Assess the perceived impact of changes from parents and their future expectations
2. Recognize the grief of parents resulted from child's body image
3. Positive reinforcement of parents to alleviate negative thoughts
4. Exhibit positivity in routine care

Nursing diagnosis: Ineffective breathing pattern related to depressed nasal bridge

Goa: Maintain an effective breathing pattern and absence of dyspnea

Interventions

1. Provide comfortable position that is sitting position to facilitate breathing.
2. Encourage for sustained deep breath.
3. Provide medication and oxygen therapy as prescribed by doctor.
4. Maintaining a clear airway.

Nursing diagnosis: Imbalance nutrition less than body requirement related to unwillingness to take meal.

Goal: To maintain the adequate weight

Interventions

1. Assess the nutritional history and etiological factors associated with decrease intake
2. Provide comfortable environment
3. Provide small and frequent meal
4. Encourage parents to have meal together

Discussion

Crouzon is one kind of rare genetic disorder caused by mutation in FGFR OR FGFR2 gene. That results from premature fusion of fibrous joints of skull. This works as an obstruction in proper growth of brain resulting alteration in the shape of skull. Severity differs from one infant to other infant. Baby X who is suffering from crouzon syndrome shown fresh mutation and has gone through invasive surgeries such as fronto-orbital advancement and craniotomy. Baby was discharged after treatment with shunt but asked for follow-up. Parents counseling was done during discharge.

References

1. Pournima G et al, Crouzon Syndrome: A Case Report. Europian Journal of Dentistry and medicine 2012;4(1):8-13.
2. Lin Yin et al (2017), C278F mutation in *FGFR2* gene causes two different types of Syndromic craniosynostosis in two Chinese patients. Molecular Medicine Report 2017 Oct;16(4):5333–37. Published online 2017 Aug 14. doi: 10.3892/mmr.2017.7248.
3. Gupta Suraj, The Short Textbook of PEDIATRICS, ed 11th 2009, Jaypee Brothers Medical Publishers (P) Ltd, P.No.
4. Kimonis V, Seminars in pediatric neurology 2007;14(3):150–61
5. Ahmed I, Afzal A, Journal of the college of physician and surgeons Pakistan 2009;19(5):318–20.
6. emedicine.medscape.com
7. Reardon W et al., Nature genetics 1994;8(1):98–103
8. http://www.childrenshospital.org/conditions-and-treatments/conditions/c/crouzon-syndrome/symptoms-and-causes

9. Jones KL, The Journal of pediatric 1975;86(1):84–8
10. Cunningham ML, Orthodontics & craniofacial research 2007;10(2):67–81
11. Roscioli T et al, American journal of medical genetics 2013;163C(4):259–70
12. Hlongwa P. et al, Journal of Maxillofacial Oral Surgery 2009;8(1):74–76
13. X Lu et al, International journal of Oral and Maxillofacial surgery 2019;48(3):309–21
14. https://rarediseases.org/rare-diseases/crouzon-syndrome/
15. Kreiborg S, Scand J PlastReconstrSurg Suppl 1981;18:1–98
16. Renier D et al. Child's nervous system 2000;16(10–11):645–58
17. Robin H Nathaniel et al. FGFR-Related Craniosynostosis Syndromes. GeneReviews® [Internet]. Initial Posting: October 20, 1998; Last Update: June 7, 2011.
18. childrenshospital.org/conditions-treatments/conditions/c/crouzon-syndrome/testing-and-diagnosis
19. Wong's; Nursing care of infants and children, ed 10th 2015, p.301, Elsevier mosby.