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SEVERE CONSTRICTED HEAD – AN EXTREME FORM OF CROUZON SYNDROME POSING CHALLENGING FRONTO-ORBITAL ADVANCEMENT: A CASE SERIES REPORTS

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ABSTRACT

Introduction: Constricted head or cloverleaf deformity in Crouzon syndrome is a severe form of the syndrome involving trilobed coronal and lambdoid synostosis. Crouzon syndrome with acanthosis nigricans is distinct from the classic Crouzon syndrome, characterized by thick and dark skin in body folds. The major problems resulting from constricted head are related to craniostenosis, orbitostenosis developing from abnormalities of the skull base associated with progressive rise in intracranial and intraorbital pressures which could progress to hydrocephalus and cerebellar herniation.
Case Illustration: Two cases with severe Crouzon syndrome were reported. The first wasa five month old girl associated with acanthosis nigricans and the other was a sixteen month old boy, both whose phenotypic expressions were at the extreme severe end of the disease spectrum. On examination there was serious corneal exposure, visual loss, severely narrow head, midface hypoplasia, and beaked nose. CT scan showed typical cloverleaf skull, expanded middle cranial fossa, foreshortened anterior and posterior fossae and honeycomb appearance in the occiput involving pansutural synostosis and extensive copper beaten deformity from the high intracranial pressure . Both were complicated with hydrocephalus requiring ventricul operitoneal shunt.
Discussion: This required an arduous effort in releasing the brain from the multiply punctured calvaria, avoiding excessive dural tear and bleeding and ultimately preserving the brain. Excessive bleeding was also caused by the raised ICP. Both patients are planned to undergo occipital expansion three months later.
Conclusion: Severely constricted head in Crouzon syndrome is an extreme manifestation and late stage of the syndrome which not only will result in irreversible complication but will require meticulous surgical technique.

Highlights:

1. Severe Constricted Head in Crouzon Syndrome.
2. Complex surgical interventions to address severe craniofacial abnormalities in Crouzon syndrome.

INTRODUCTION

Modern era of craniostenosis surgical treatment began in 1970, when plastic surgeons and neurosurgeons joined forces to form craniofacial teams and described new techniques to solve thefunctional and the structural problems at the same time. The techniques developed at the Hopital des Enfants Malades in Paris were published in detail in 1982, and although

some minor changes have been introduced the basic techiques remain unchanged after treatment of over 1,600 patients[3].

Crouzon Syndrome is among the rare syndromic craniosynostosis inherited by autosomal dominant transmission, affecting cranial development in consequence of formation from two or more suturepremature fusion called craniofacial dysostosis. This is characterized by craniosynostosis, exophthalmos, and midface hypoplasia. The birth prevalence of Crouzon syndrome is estimated to be 15–16 per one million births[3].

The majority of patients with Crouzon syndrome have mutations in the extracellular immunoglobulin III domain of the fibroblast growth-factor receptor 2 (FGFR2) gene.

Crouzon syndrome with acanthosis nigricans is rare; this condition occurs in about 1 person per million. For unknown reasons, it affects females more than twice as often as males. The genetic change involved in this disorder causes the FGFR3 protein to be overly active, which disrupts the normal growth of skull bones and affects skin pigmentation[10]. The main point in Crouzonsyndrome is that the synostosis is frequently progressive. Usually, the sutural fusions do not exist at birth. The coronal and sagittal fusions appear at about one year of age, the lambdoid later in life[5].

The craniofacial deformity depends on which sutures are affected. Constricted head deformity in Crouzon syndrome is a severe form of the syndrome which may be a consequence of multiple sutural fusion, due toa constriction ring developing in the lambdoid-squamosal zone, causing bulging in the frontal and temporal bones and a trilobar head shape[8].

This constricted head has other names such as cloverleaf skull (CLS), kleeblatshadel and triphylIocephaly9. The main features are cloverleaf deformity characterized by facial hypoplasia involving the orbits causing usually severe exophthalmos, and the nose and upper maxilla and occasionally severe hydrocephalus and poor prognosis due to progressive intracranial hypertension5. The forehead is recessed vertically, with retrusionof the supraorbital rim and root of the nose[3]. A review article by Cinalli (2005) reported that Chiari I malformation was found in 100% of cloverleaf skull deformity12. Management of CLS in Crouzon syndrome follows the same established protocols as for other craniosynostosis. Two universally accepted protocols are those from ACFU and Erasmus CF center12.

CASE ILLUSTRATION

Presented two cases of severe Crouzon syndrome with constricted head, a five month female baby and a sixteen month male baby who presented to Dr.Soetomo general hospital referred from the general hospitalsin their area.

Their chief complaints were progressively abnormal head shapes since birth, difficulty in breathing, inflammation of both eyes, and progressively rapid visual loss with protruding eyes. They always snored and would wake up suddenly in the night apparently from difficult breathing. Perinatal history was unremarkable. Family history revealed no craniofacial deformity.

Case 1

A 6 month old female, admitted to hospital with abnormal head shape from birth. She is the second child from her family with normal aterm birth, weight 3300 g, length 49 cm, with normal limbs.

Examination of the head the baby showedsevere constricted head lover leaf

or trilobular deformity with bulging at the anterior fontanalle like oxycephaly associated with bilateral temporal bulges. brachicephaly, turricephaly, midface hypoplasia that make a baked nose, with cephalic index 13.3 and head circumference 38 cm

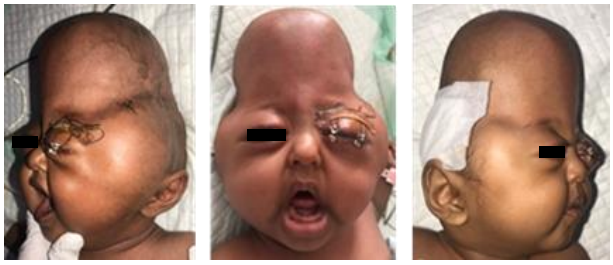

Figure 1. Craniofacialis region from 5 month female patient

There was bilateral exorbitism with the left eye being very proptotic with constant epiphora since two weeks before she came to Dr.Soetomo general academic hospital. There waÓ bilateral lagophtalmus, corneal ulcer on the left eye, and exposure keratitis with papil atrophy on the right eye. From ultrasonography examination there was no sign of intraocular infection, and normal vitreoretinal. ENT finding. This patient snored during sleep with nasal congestion ×iôÈÏÕÔ a ÈiÓÔÏÒÙ aÐÎeaß 4ÈiÓ ÐaÔieÎÔ Èad deÌaÙed ÇÌÏÂaÌ deÖeÌÏÐmeÎÔaÌ ÐÒÏÂÌem iÎ ÂÏÔÈ ÐÙÓcÈÏmÏÔÏÒ aÎd ÐÓÙcÈÏÓÏciaÌß %ØÔÒemiÔieÓ ×eÒe ÎÏÒmaÌiÎ ÆÕÎcÔiÏÎ aÎd mÏÒÐÈÏÌÏÇÙß ! #4 ÓcaÎ ÏÆÔÈe Èead ed ÈÙdÒÏceÐÈaÌÕÓ ÎÏÎ cÏmmÕÎicaÎÓℏ ÓÙÎÏÓÔÏÓiÓ ÏÆÔÈe meÔÏÐicℏ ÂiÌaÔeÒaÌ cÏÒÏÎaÌ aÎd ÌamÂdÏid ÓÕÔÕÒeÓ ×iÔÈ ÓeÖeÒe cÏÐÐeÒ ÂeaÔeÎ aÐÐeaÒaÎceß

This patient had eye infection resulting from severe exorbitism. Surgery had to be postponed until the infection was healed. While awaiting surgery his left eye was protected with a tarsorrhaphy. Soon after arriving at the ER, the neurosurgeon performed a emergency ventricul operitoneal shunt in which the dissection was difficult due to the entrapped dura in the multiple holes on the thin calvaria causing extensive bleeding. Because long durante and difficult operation cause a lot of 1500 ml bleeding.

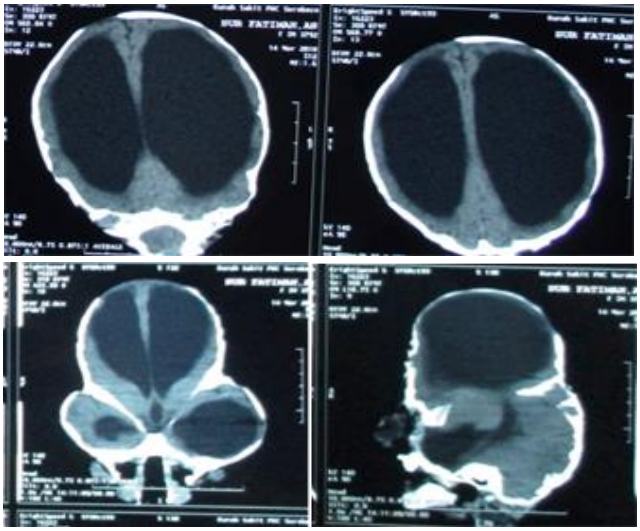

Figure 2. CT-Scan showed cloverleaf deformity and hydrocephalus

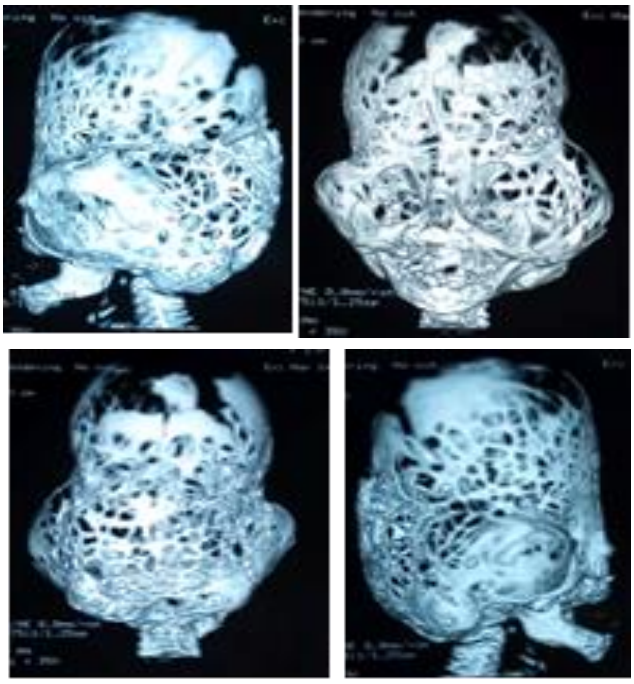

Figure 3. Cranial 3D reconstruction CT-Scan showed copper beaten apearance

It was decided to do a craniotomy/suturectomy, fronto-orbital advancement (FOA) and cranial vault remodelling with the neurosurgical team after splitting the palate to secure the airway after extubation.

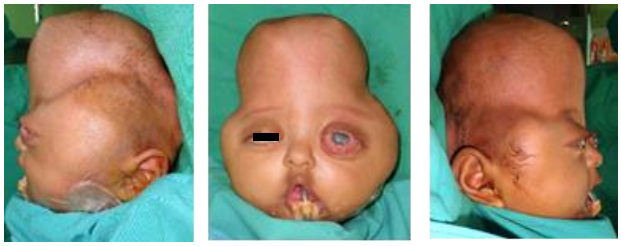

Figure 4. Before fronto-orbital advancement

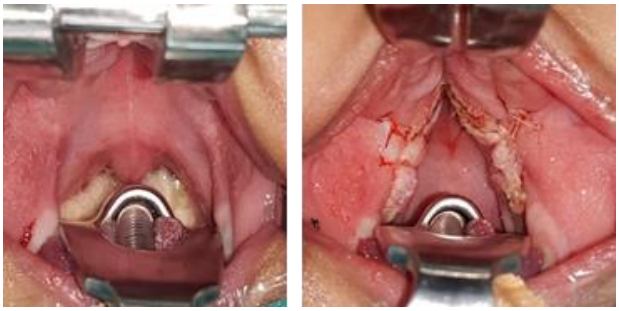

Figure 5. Before and after palatal split, fixing the soft palate to the alveolus with 4.0 silk suture.

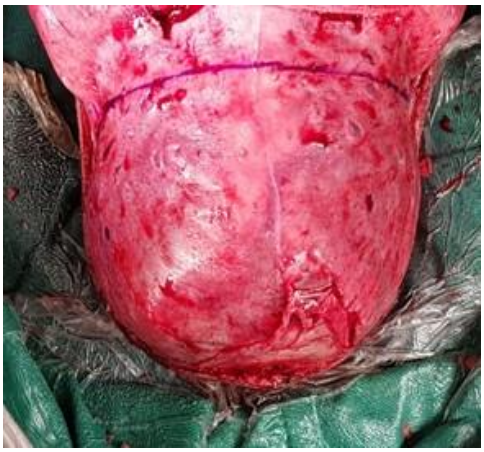

Figure 6. Showed hole in duramater

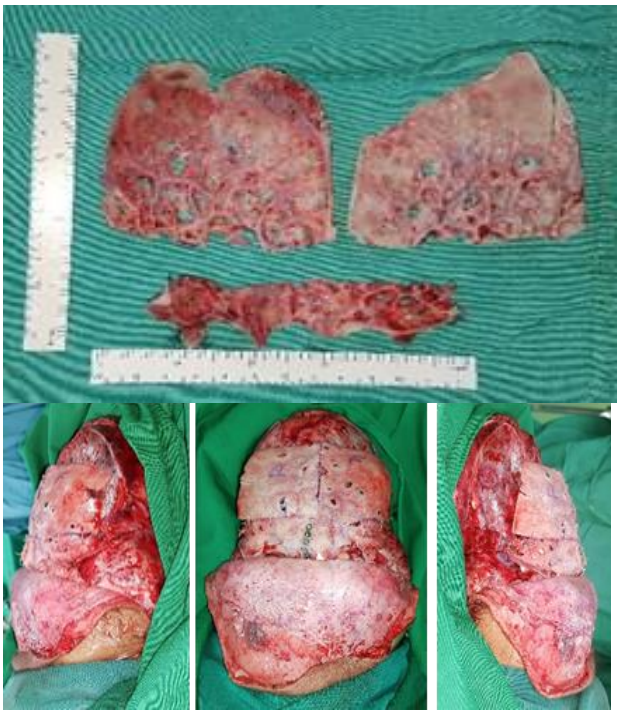

Figure 7. Durante Operation, 1 cm fronto-orbital advancement

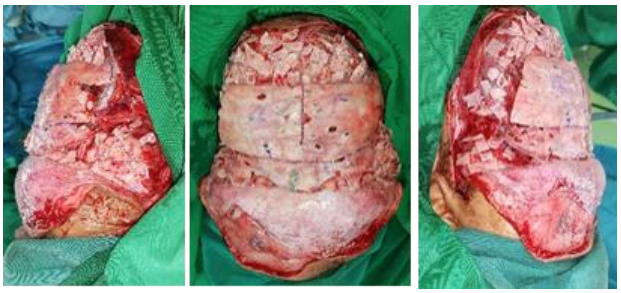

Figure 8. Filling the remaining cranial gaps with fibrin glue and bone chips

Figure 9. After fronto-orbital advancement

Case 2

A-16 month old male patient was

admitted to hospital with abnormal head shape and eyeball protrusion since seven month old before. He is the last child from his family birth atterm with section secaria, weight 3300 g, length 50 cm, with normal limbs. Examination of the head the baby showed severe constricted head clover leaf deformity, brachicephaly, turricephaly, trigonocephaly, with ridging on both coronal, sagital sutures, and both lambdoid sutures with head circumference 47 cm.

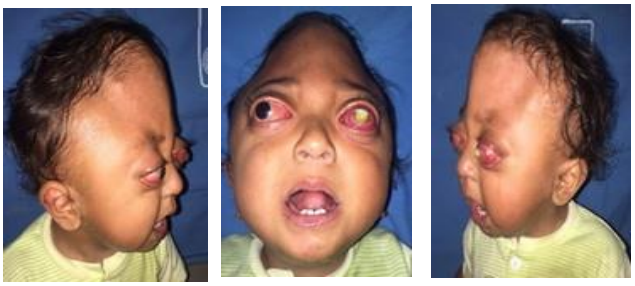

Figure 10. Preoperative appearance

Both eyes were displaced forward whenever baby cried since seven months old. But in thirteen month age, the protrusion became more fixed . There are bilateral lagophtalmus, bilateral proptosis with corneal ulcer on the left eye, and exposure keratitis on the right eye. Intra-oral examination revealed high arched palate, hypoplastic maxilla and chronic adenotonsilitis. Polysomnography revealed mixed moderate central and obstructive sleep apnea. This patient had delayed global developmental problem in both pyschomotor and psychosocial. All extremities were normal in function and morphology. CT scan showed Kleeblattschadel head deformity with premature closure of sagittal, bilateral coronal, frontal, both lambdoid sutures. Right and left lateral system ventricle III-IV were very dilated indicating a communicating hydrocephalus. The exophthalmos on the right eye protruded 1.16cm and the left eye 1.19 cm.

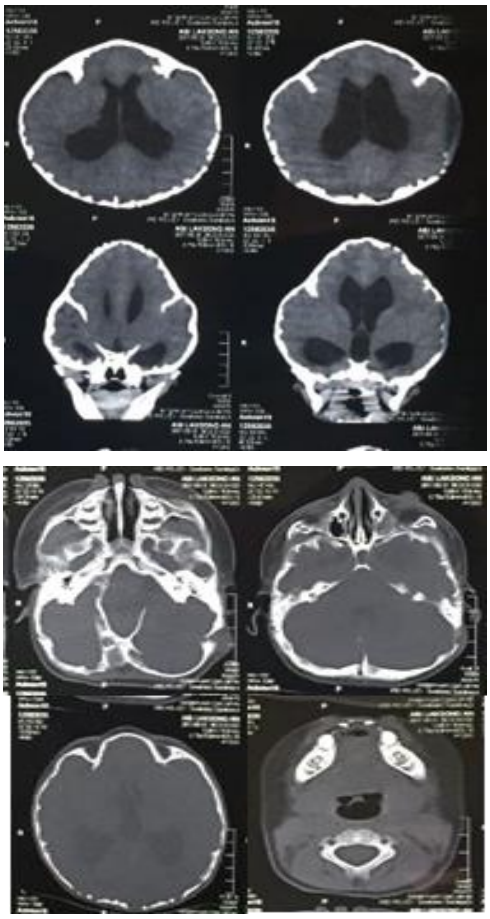

Figure 11. Bone window on computed tomography scanFigure 14. Cranium CT

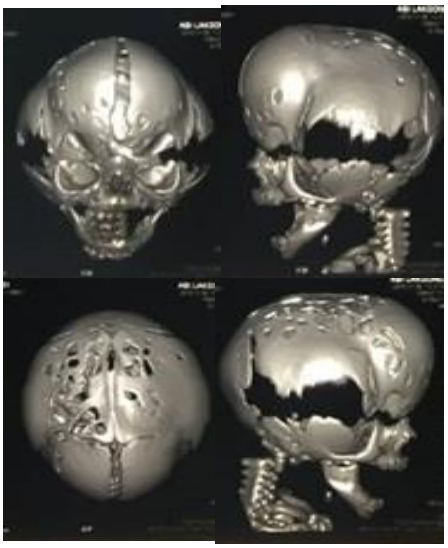

Figure 12. Cranium CT performed; 9

month after fronto-orbital advancement operation

Figure 13. Before operative fronto-orbital advancement

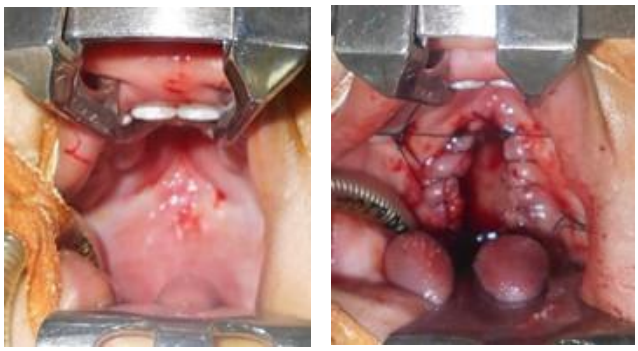

Figure 14. Before and after palatal split, fixing thesoft palate to the alveolus with 4.0 silk suture.

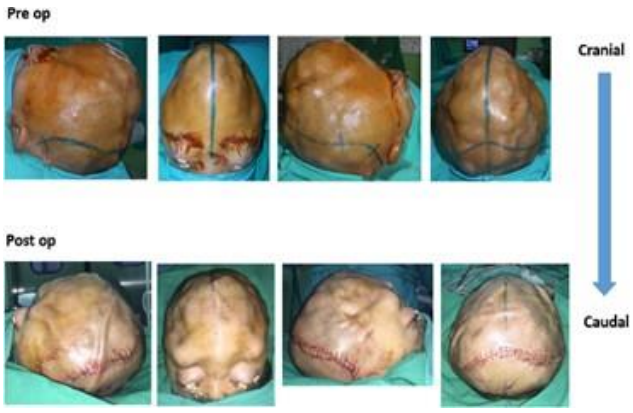

Figure 15. Pre and post fronto-orbital advancement procedure, showed temporo parietal occipital region

Difficulties from this patient are high intracranial pressure caused by hydro-cephalus communicans and make a copper beaten appearance. Difficult airway caused obstructive sleep apnea make a challenge to anesthesiologist. Estimation blood lost of this surgery is 180 ml. It was decided to do a split palate to secure the airway after extubation then a craniotomy/ suturectomy, fronto-orbital advancement (FOA), cranial vault remodelling and tapping the ventricle with the neuro-surgical team.

DISCUSSION

Constricted head in the form of CLS or kleeblatshadel can be found in several craniosynostosis syndrome including Crouzon syndrome, Boston type and and Thanatophoric dysplasia type II[11].

Table 1. Clover leaf skull deformity[11].

No.	Condition	Craniofacial Phenotype	Gene
1.	Boston Type	Craniosynostosis,cloverleaf skull,forehead retrusion, frontalboosing.	*MSX2*
2.	Thanatop-horic dysplasia type II	Craniosynostosis,cloverleaf skull, severe bonegrowth disturbance	FGFR3

Both cases were diagnosed with Crouzon syndrome based on clinical findings. Both has CLS which is a severe form of Crouzon syndrome with serious complications including compromised airway and breathing, increased intracranial pressure, visual loss and ulceration. These were neglected cases because of delay in diagnosis and subsequently in treatment.

This condition was supposed to have been managed by standard protocols. In both patients, suturectomy, FOA and cranial vault remodeling were done to

correct fronto- supraorbital retrusion, to protect the globes and to expand the intracranial volume.

The underlying pathology of a CLS is a constriction ring in the lambdoid-squamosal zone[8].

Table 2. Standard protocol from Erasmus CF center[12].

Australian Craniofacial Unit	
Birth – 3 months	**Total Assessment**
3 – 6 months	Planning meeting Surgery : - Fronto-orbitalAdvancement - Bone Graft - Lambdoid Craniectomy
1 year old	- Total review - Craniofacial clinic
1 – 10 years old	- Yearly review and craniofacial clinics 3 yearly - Dentistry 6 monthly checks - Orthodontictreatment Surgery : - Facial/Fronto-facial Advancement - Bone graft
Teenage years	- Complete Workup - Dentistry 6 monthlychecks - Orthodontictreatment - Orthognathic surgery
Maturity	Touch-up surgery
Erasmus Medical Center Rotterdam	
Pre operative CT, MRI, Funduscopy, Sleep Study	
6 months	- Occipital expansionwith springs - Endoscopy of upperairway - If luxatingeyeball/severe OSA : Early Monobloc
8– 12 years or >18 years old	Monobloc or Facial Bipartition
18 years old	Le Fort I ± BSSO

Fusion of lambdoid sutures often result in significant posterior cranial fossa volume, narrowing of central nervous system and associated with hydrocephalus which also be caused by venous hypertention[13].

The cystic dilatation of the temporal horns is the earliest sign of hydrocephalus and occurs secondary to disturbed growth of the cerebral capsule 9. In both patients, there was hydrocephalus. One had to be treated with emergency VP shunt before cranial expansion and the other with tapping of the ventricle at the same time with cranial expansion.

The skull X-ray showed typical CLS with expanded middle cranial fossa, foreshortened anterior and posterior fossae and honey-comb appearance in the occiput. CLS often involves lambdoid and squamosal sutures[9].

On the head CT scans of both patients, there were the synostosis involved almost all the cranial sutures including the lambdoid and squamosal sutures[9]. In both patients, dueto the chronic and significantly increased ICP, complicated with hydrocephalus, the pressure upon the internal table of the skull became so great, meticulous and arduous dissection had to be carried out to avoid dural tear and bleeding. Excessive bleeding was also caused by the raised ICP. The mechanismof ventricular dilation in Crouzon syndrome is not clear , and there are various theories. It could be caused by constriction of the subarachnoid spaces, or it could be due to to obstruction of venous drainage from venouse sinuses, as suggested by Renier et al[6].

Acanthosis nigricans is a skin condition characterized by thick, dark, velvety skin in body folds and creases, including the neck and underarms[10].

CONCLUSION

Severely constricted head is an extreme manifestation and late stage of Crouzon syndrome. This could have been prevented by early diagnosis and multidisciplinary management according to established protocol.

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CONFLICT OF INTEREST

There is no conflict of interest in this article.

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The authors herely declare they have no financial interest in the information discussed in this article.

AUTHORS CONTRIBUTION

All authors contributed to the conceptualization, data interpretation, methodology, manuscript writing, and revised the manusript.

REFERENCES

1. Glaser RL, et al. Paternal origin of FGFR2 mutations in sporadic cases of Crouzon syndrome and Pfeiffer syndrome. Am J Hum Genet 2000;66:768-77.
2. Granger B., et al. Analysis of Fronto-orbital Advancement for Apert, Crouzon, Pfeiffer, and Saethre-Chotzen Syndromes, 2000.
3. Renier D., et al. Management of craniosynostoses, 2000.
4. Ghassan S., et al. Raised Intracranial pressure in crouzon syndrome: incidence,causer,and management., 2016.
5. Iannaccone G., Gerlini G., The So-called *Cloverleaf Skull Syndrome", 1974.
6. Hanieh A., et al. Hydrocephalus in Crouzon's syndrome, 1989.
7. Granger B., et al. Analysis of Fronto-orbital Advancment for Apert, Crouzon, Pfeiffer, and Saethre-Chotzen Syndromes, 2000.
8. David JD, et al. The Craniosynostoses: Causes, Natural History, and Management. Springer-Verlag London. 1982.
9. Rohatgi M, Cloverleaf Skull – a Severe Form of Crouzon's Syndrome : a New Concept in Aetiology ,1991.
10. Hill.J, Lister Hill National Center For Biomedical Communication, Crouzon Syndrome With Acanthosis Nigricans, 2018.
11. Guyuron R., et al. Plastic Surgery Indcations and Practice, Saunders Elsevier, 2009.
12. Irene, Guidline Treatment and Management of Craniosynostosis, The netherland Society for Plastic Surgery. 2010.
13. Machado G, et al. Cloverleaf skull deformity and hydrocephalus. Childs Nerv Syst.2011.27:1683–1691.