Craniosynostosis: an ocular perspective

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Premature fusion of one or more cranial sutures leading to abnormal head shape.

Classified asprimary and secondary. Also as simple , complex and syndromic Includes :

a. Crouzon syndrome

b. Apert syndrome

c. Pleiffer syndrome

d. Carpenter syndrome

Ossification of cranial vault starts in the central region of each cranial bones,then extends towards sutures.

- Primary-when one or more suture fuse prematurelyprimary ossification. Defect. e.g. scaphocephaly, brachycephaly, plagiocephaly, trigonocephaly.
- Secondary - primary failure of brain growth, more common, 90% of total

Baller-Gerold Syndrome

Ocular Features

The ocular features are a rather minor part of this syndrome and are found in less than a third of patients. These primarily involve lids and adnexae with telecanthus, downslanting lid fissures, and epicanthal folds. Some also have nystagmus while strabismus, blue sclerae, and ectropion have also been reported.

Systemic Features

The cardinal features of this syndrome are craniosynostosis and radial defects. However, a large number of variable defects such as imperforate or anteriorly placed anus, rectovaginal fistula, absent thumbs, polydactyly, and mental retardation may also be present. The radius may be completely absent or abnormally formed and occasionally the ulnar bone is involved as well. Some patients have a conductive hearing loss.

Treatment Options

No treatment is available.

References

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2. Temtamy SA, Aglan MS, Nemat A, Eid M. Expanding the phenotypic spectrum of the Baller-Gerold syndrome. Genet Couns. 2003;14(3):299-312.

Carpenter Syndrome

Ocular Features

A variety of ocular anomalies have been reported in Carpenter syndrome with none being constant or characteristic. The inner canthi are often spaced widely apart and many have epicanthal folds and a flat nasal bridge. Other reported abnormalities are nystagmus, fovealhypoplasia, corneal malformations including microcornea, corneal opacity, and mild optic atrophy and features of pseudopapilledema.

Systemic Features

Premature synostosis involves numerous cranial sutures with the sagittal suture commonly involved causing acrocephaly (tower skull). Asymmetry of the skull and a 'cloverleaf' deformity are often present. The polydactyly is preaxial and some degree of syndactyly is common especially in the toes. The digits are often short and may be missing phalanges. Some patients are short in stature. Structural brain defects may be widespread including atrophy of the cortex and cerebellarvermis. Septal defects in

the heart are found in about one-third of patients. The ears can be low-set and preauricular pits may be seen. Some but not all patients have obesity and a degree of mental retardation.

Treatment Options

No treatment of the ocular defects is necessary in most cases. Craniectomy may be required in cases with severe synostosis.

References

1. Hidestrand P, Vasconez H, Cottrill C. Carpenter syndrome. J Craniofac Surg. 2009 Jan;20(1):254-6.
2. Jenkins D, Seelow D, Jehee FS, Perlyn CA, Alonso LG, Bueno DF, Donnai D, Josifova D, Mathijssen IM, Morton JE, Orstavik KH, Sweeney E, Wall SA, Marsh JL, Nurnberg P, Passos-Bueno MR, Wilkie AO. RAB23 mutations in Carpenter syndrome imply an unexpected role for hedgehog signaling in cranial-suture development and obesity. Am J Hum Genet. 2007 Jun;80(6):1162-70.
3. Temtamy SA. Carpenter's syndrome: acrocephalopolysyndactyly. An autosomal recessive syndrome. J Pediatr. 1966 Jul;69(1):111-20.

Pfeiffer Syndrome

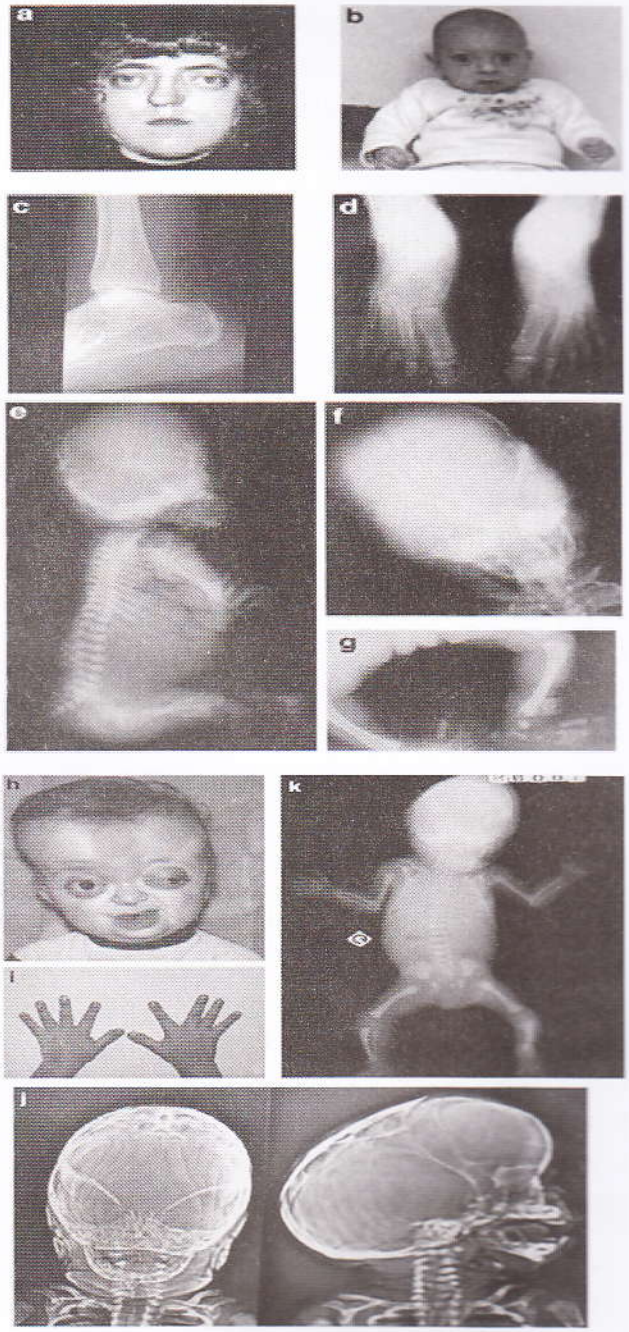
Ocular Features

Patients may have extreme proptosis secondary to shallow orbits and exposure keratitis is a risk. Hypertelorism, strabismus, and antimongoloid lid slants are common. More rare signs include anterior chamber anomalies and optic nerve hypoplasia.

Systemic Features

Pfeiffer syndrome has been divided into 3 types, of which cases with types 2 and 3 often die young. Type 1 has the more typical features with midfacehypoplasia, broad thumbs and toes, craniosynostosis, and often some degree of syndactyly. Adult patients with type 1 may be only mildly affected with some degree of midfacehypoplasia and minor broadening of the first digits. Hearing loss secondary to bony defects is relatively common. Cleft palate is uncommon. Airway malformations especially in the trachea can cause respiratory problems.

Treatment Options



Exposure keratitis requires the usual treatment. Surgery for the midface underdevelopment may be helpful for the proptosis. Airway obstruction may require tracheostomy or surgical correction of the air passages.

References

1. Glaser RL, Jiang W, Boyadjiev SA, Tran AK, Zachary AA, Van Maldergem L, Johnson D, Walsh S, Oldridge M, Wall SA, Wilkie AO, Jabs EW. Paternal origin of FGFR2 mutations in sporadic cases of Crouzon syndrome and Pfeiffer syndrome. Am J Hum Genet. 2000 Mar;66(3):768-77.

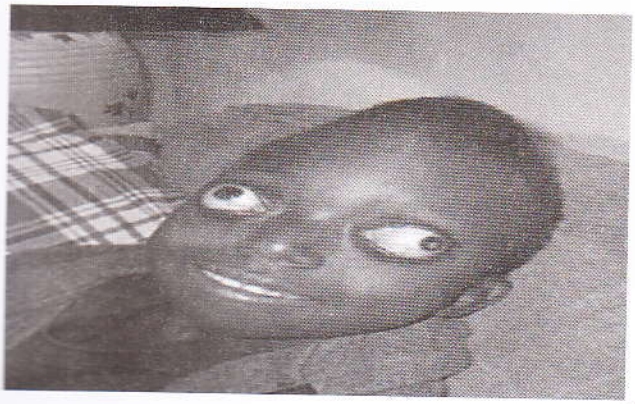
2. Schell U, Hehr A, Feldman GJ, Robin NH, Zackai EH, de Die-Smulders C, Viskochil DH, Stewart JM, Wolff G, Ohashi H, et al. Mutations in FGFR1 and FGFR2 cause familial and sporadic Pfeiffer syndrome. Hum Mol Genet. 1995 Mar;4(3):323-8.

3. Lajeunie E, Ma HW, Bonaventure J, Munnich A, Le Merrer M, Renier D. FGFR2 mutations in Pfeiffer syndrome. Nat Genet. 1995 Feb;9(2):108.

Crouzon Syndrome

Ocular Features

The primary ocular features result from pattern-specific, premature synostoses of cranial sutures. The orbits are often shallow resulting in proptosis, sometimes to such an extent that exposure keratitis or even spontaneous subluxation of the globe results. This is exacerbated by the midfacehypoplasia that is often present. As many as 22% of patients have optic atrophy, most likely secondary to chronic papilledema from elevated intracranial pressure. Strabismus is common, often with a V-pattern exotropia. Overaction of the inferior obliques and underaction of the superior obliques have been described.



Systemic Features

The coronal sutures are the most commonly affected by the premature synostosis and hence the skull is often brachycephalic and the forehead is prominent. Increased intracranial pressure is a risk. The nose is parrot-beaked and the upper lid is short. Maxillary hypoplasia from the midface underdevelopment can cause crowding and displacement of the upper teeth.

Treatment Options

Exposure keratitis must be treated. Cranial surgery has been necessary for some patients to relieve the papilledema but the post operative outcome can be complicated by hydrocephalus.

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Bertelson TI. The premature synostosis of the cranial sutures.ActaOphthalmol Suppl. 1958;36(Suppl 51) : 1-176.

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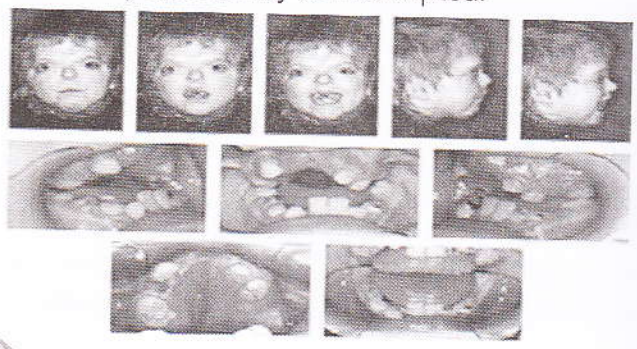
Apert Syndrome

Ocular Features

In 10% of patients, keratitis and corneal scarring occur from the sometimes marked proptosis and corneal exposure. Optic atrophy is present in over 20% of patients. Strabismus, primarily exotropia, is found in more than 70% and various extraocular muscle anomalies may be detectable. Usually the exotropia has a V-pattern with overaction of the inferior oblique muscles while the superior oblique is weak. Amblyopia occurs in nearly 20%. The lid fissures often slant downward and the eyebrows may be interrupted.



Systemic Features

This brachysphenocephalic type of acrocephaly is associated with syndactyly in the hands and feet. Pre- and postaxial polydactyly may be present. There is considerably variation in expression with some patients so mildly affected that they appear virtually normal, whereas others have extreme degrees of brachycephaly with high foreheads, midfacehypoplasia, and proptosis secondary to shallow orbits. Imaging often reveals one or more CNA anomalies such as defects of the corpus callosum, partial absence of the septum pellucidum, ventriculomegaly, and sometimes hydrocephalus. A small but significant proportion of patients have some developmental delay and cognitive impairment. Over 39% of patients have a normal IQ.

Treatment Options

No specific treatment is available for this disorder but exposure keratitis may require surveillance and therapy.

References

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- *The human eye can distinguish about 10 million different colors.*
- *Some women can have a genetic mutation which causes them to see millions of more colors.*
- *People with blue eyes have a higher alcohol tolerance.*
- *If the human eye was a digital camera it would have 576 megapixels.*
- *All blue eyed people can be traced back to one person who lived near the Black Sea almost 10,000 years ago.*
- *We spend about 10% of our waking hours with our eyes closed, blinking.*
- *Researchers have successfully used the game TETRIS to treat "lazy eye" in adults.*
- *Albert Einstein's eyes remain in a safe box in NYC.*
- *Your eyes can get sunburned.*
- *Black lemurs are thought to be the only primates, besides humans, to have blue eyes.*
- *The space between your eyebrows is called Nasion.*