Vejovis Journal

APERT SYNDROME – A Rare Encountered Clinical Entity. A case report

Mr M. A. Calcuttawala MD[1], Dr Mohit Bhatia[2]

[1] Upper Gastrointestinal Surgery, Newham University Hospital, Glen Road, E138SL London UK
[2] Department of General Surgery Kings College Hospital, Denmark Hill, SE5 9RS, London, UK

Abstract

It is a complex syndrome entity which is less commonly seen and due to its multisystem involvement and its complexity rendering the optimum treatment remains a surgical challenge. This syndrome is characterised by facial deformities, and irregular bony growth patterns. Due to delay in making a proper diagnosis and framing the treatment patterns, even after surgical interventions results are mostly not as desired. We present a case of a 9 year old male patient who presented to us with this complex entity.

Introduction

It is a rare congenital disease which is characterised by facial deformities along with improper fusion of skull bones (craniosynostosis) due to which intracranial pressure is believed to increase and hence causing more deformities. [1]

It is an autosomal dominant condition which involves syndactyly of hands and feet. [2] In 1906, Eugene Apert first presented this condition which is believed to have an incidence of 1 in 100,000 to 1:150,000. [3]

This syndrome is believed to occur due to mutation at residues Ser 252 Trp and Pro253 Arg. [4] Fibroblasts growth factors are polypeptide ligands which play a role during foetal and embryonic life. Mutations in FGF receptors results in craniosynostosis.[5] Craniosynsotosis can be a feature of various other syndromes like crouzon syndrome.[6] Crouzon syndrome never involves limbs where as in aperts syndrome there occurs inappropriate fusion of bones of hands and feet. [7]

Apert syndrome is classified as a form of acrocephalosyndactyly which includes malformation of skull and webbing of fingers of hands and toes. [8 Apert

syndrome also affects oral cavity which is characterised by malformations which includes crowding of teeth, disfigured maxilla, and ectopic teeth eruptions. However mostly mandible is of normal size and shape, but because of choanal stenosis and ill formed palate patients often presents with mouth breathing. [9]

Case report

A 9 year old male patient presented to us with features of dysmorphic facial features , ocular deformities with fused digits of both hands and feet.(Fig 1,2,3) On examination we found patient having irregular dentition and altered speech. Patient had protuberant eyes, depressed nasal bridge. (Fig 1) Detailed history did not reveal anything substantial. His mother had no history of any prolonged medication. He had 1 sibling who was completely normal. Patient had normal intelligence and showed no features of mental retardation and had normal developmental milestone history. Patient had undergone surgery for craniosynostosis at age of 4 years however there was no documentation provided. CT scan head was suggestive of non visualisation of corpus callosum, with large bony gap in midline involving bilateral frontal bones with partial fusion of saggital suture with complete fusion of coronal and lambdoid sutures with non visualisation of sutural markings with findings suggestive of craniostenosis with agenesis of corpus callosum.

Discussion

It is a rare encountered clinical condition. It is an autosomal dominant condition which comprises mainly of dysmorphic facial features along with syndactyly of hands/feet along with craniosynostosis.[10] It is believed mutations of genes located on chromosome 10 are responsible for resulting in premature fusion of coronal and saggital sutures. Early fusion of these sutures are often associated with optic atrophy, exopthalmos and altered dentition. [11]

Mutations of fibroblasts growth factors results in events which ultimately affects the cell division and differentiation. Severity of signs and symptoms are influenced by the variety of mutations. In a study by Slaney he concluded that Pro253Arg mutations are often associated with the most severe forms of Apert syndrome.[12]

Incidence of Apert syndrome increases with the paternal age, and its incidence is more in males. It can be diagnosed with

the help of ultrasonography during the antenatal period. This syndrome is often confused with the crouzon syndrome due to similar features. Although in apert syndrome there is midline defect seen unlike crouzon syndrome. Patient's needs to be treated at an early age for limb defects as the quality of life and disability index often depends on the outcome of the surgical interventions. [13]

Conclusion

In modern era with multimodal approach to diagnose medical problems at an early stage we are still missing on finding the right direction to diagnose the less heard medical conditions which are equally hazardous for the patients. We need to have a multidirectional approach to deal with such less known conditions, as optimum treatment can improve the quality of life for the patient and can enable them to have a life without emotional and physical liability.

Conflict of Interests

The authors' declare that there are no conflicts of interests.

Acknowledgments: None

Funding information was not available.

Keywords: Syndactyly, crouzon syndrome, craniosynostosis, apert.

Correspondence:

Mr M. A. Calcuttawala MD

Newham University Hospital, Glen Road, E138SL London UK

murtuza.calcuttawala@nhs.net

References

1. Varoli FP, Santos KCP , Costa C , Olievera JX. Apert syndrome : clinical and radiographic features and case report. Rev Odonto Cienc 2011;26(1):96-99.
2. Verma S, Draznin M Apert syndrome. Dermatology Online Journal 2005;11(1):15.
3. Sannomiya EK, Reis SAB, Asaumi J, Silva JVL, Barbara AS and Kishi K. Case report. Clinical and radiographic presentation and prepration of the prototyping model for presurgical planning in Aperts syndrome. Dentomaxillofacial Radiology 2006;35:119-124.
4. Wilkie, A.O.M., Slaney, S.F.,Oldrige, M.,Poole, M.D.,Ashworth, G.J Hockley et al. Apert syndrome results from localised mutations of FGFR2 and is allelic with crouzon syndrome. Nature genet.,9,165-172.
5. Muenke,M.,Schell,U.,Hehr,A.,Robin,N.H. ,Losken,H.W.,Schinzel,A.,Pulleyn,L.J.,Rutland,P.,Reardon,W.,Malcolm,S. and Winter,R.M.(1994) Mutations in the fibroblast growth factor receptor-1 gene in Pfeiffer syndrome. Nature Genet.,8,275-279.
6. Reardon,W.,Winter,R.M.,Rutland ,P.,Pulleyn,L.J.,Jones,B.M. and Malcolm,S.(1994)Mutations in the fibroblast growth factor receptor 2 gene

cause crouzon syndrome. Nature Genet., 8, 98-103.

7. Wilkie,A.O.M.(1997) Craniosynostosis :genes and mechanisms.Hum.Mol.Genet.,6,1647-1656.

8. Fearon, Jeffrey A. Treatment of the hands and feet in apert syndrome.: An evolution in management." Plastic and reconstructive surgery. 112(1):1-12(2003)

9. Frieman A, Tessler O, Barahnin B. Apert syndrome. Int J Dermatol 2006;45:1341-3.

10. DeGiovanni CV, Jong C, Woollons A. What syndrome is this? Apert syndrome. Pediatr Dermatol 2007;24:186-8.

11. Cohen MM Jr, Kreiborg S. The growth pattern in Apert syndrome. Am J Med Genet 1993;45:617-623.

12. Slaney SF, Oldridge M. Hurst JA et al. Differntial effects of FGFR2 mutations on syndactyly and cleft palate in apert syndrome. Am J Hum Genet 58:923-32,1996.

13. Panchal J, Uttekin V. Management of craniosynsotosis