Pierre Robbins Syndrome

M. Imran*, M.B;B.S., F.A.A.P., D.C.H., (Lond.), D.C.H. (Dublin), Dip. Amer. Board of Paed.
Faiz M. Khan**, M.B;B.S. D.C.H. (Ireland), M.R.C.P.(Paeds. U.K.)
and
Sajjad-ur-Rehman***, M.B;B.S.
Postgraduate Medical Institute,
Lady Reading Hospital,
Peshawar, Pakistan

Summary

Pierre Robbins Syndrome, a not uncommon congenital anomaly, is diagnosed by the clinical triad of micrognathia, glossoptosis and cleft palate. When associated with other syndromes, it is called Robbin's anomaled. Certain associated anomalies in our patient have not previously been described with the syndrome. Management of infants with this syndrome, though difficult and painstaking, is nontheless rewarding.

Introduction

Pierre Robbins Syndrome has a prevalence of 1:30,000 live births[1]. It is a congenital anomaly, probably resulting from persistent intrauterine malpositioning of the fetus. It presents with its characteristic triad at birth[2,3] and feeding problems are the commonest complaints[2]. The prognosis is excellent, provided the baby is managed properly, which in fact is a challenging job for the nursery staff.

* Professor and Head, Department of Paediatrics, Postgraduate Medical Institute and Consultant Paediatrician, Lady Reading Hospital.
** Assistant Professor of Paediatrics, Postgraduate Medical Institute and Consultant Paediatrician, Lady Reading Hospital.
*** F.C.P.S.-II (Paeds) Trainee Registrar, Postgraduate Medical Institure, Lady Reading Hospital.

Case Report

A baby borne at home, to Mrs. T., a multi-gravida 30 year old mother, after full term pregnancy and normal vaginal delivery, was brought to Newborn Nursery, LRH on 11-4-1987, four hours after its birth. The parents complained that the baby was unable to take feeds, had an abormally small chin and malshaped feet. The perinatal history was suggestive that the baby had persistent transverse lie till late in pregnancy. The foetal movement started late in the 7th month of intra-uterine life.

On examination, the baby was found to have micrognathia, hypertelorism with antimongoloid eyes, cleft palate, low set ears, rocker-bottom feet with abnormal fanning of toes. Left foot had four digits. The baby was in respiratory distress and had rales bilaterally in the chest. Chest X-ray was normal and abdominal ultrasound did not reveal any other abnormality. A clinical diagnosis of Pierre Robbins Syndrome was made.

The baby was nursed in a prone position, which would markedly diminish his respiratory distress. He was given I.V. fluids, prophylactic antibiotics, bronchodilators and oxygen as needed.The chest became completely clear on the third day and he was started on gavage feeding, which he tolerated well in the prone position. Bottle feeding and supine position would produce respiratory distress. The parents were educated about proper positioning and feeding techniques and the baby was discharged home. Follow-up at one week did not show any significant clinical abnormality. Later on, the family was lost to follow-up.

Discussion

The syndrome was first described by Pierre Robbins in 1923 in his article published in Bull. Acad. Med. (Paris)[4]. Later he reviewed the syndrome in the same journal[5] and in American Journal of Diseases of Children[6]. Cohen, M.M. Jr[7,8] described other associated anomalies with P.R. syndrome and introduced the term "Robbin's Anomalad" for such cases[9]. Various associated syndromes include Stickler, Morquio, Campomelic dwarfism, Persistent left superior vena cava, Cerebro-costo-mandibular and Spondylo-epiphyseal dysplasia congenita[7].

Isolated Pierre Robbins Syndrome is not inherited[3] and most cases are sporadic[2]. When present with other syndromes, the inheritence is that of the other syndrome[3].

The etiology of the syndrome is unknown, but the most likely

hypothesis is that a persistent abnormal foetal position with marked flexion of the neck prevents forward growth of the mandible. This results in retention of tongue in nasopharynx between the palatal shelves, which fail to unite and thus cleft palate results. A decreased intrinsic growth potential of the mandible also pays a part in the genesis of micrognathia in P.R. Syndrome[9].

The classical triad essential for the diagnosis of P.R. Syndrome is micrognathia, glossoptosis and high arched/cleft palate, found in 40-70% of cases[9]. Other occassional findings are cataract, microphthalmos, glaucoma, retinal detachment, congenital heart disease, low set ears, mental retardation, hydrocephalus, microcephaly, myopia, esotropia, sixth nerve palsy, Mobius syndrome, spina bifida, hip dislocation, syndactylia and club foot[1,2]. Various anomalies of middle and internal ears and conductive hearing loss have also been described.[1]

Our patient had some associated anomalies like Rocker bottom feet, four toes in left foot and antimongoloid eyes, which to our knowledge have not been described in association with P.R. Syndrome. Hypertelorism is present in Stickeler syndrome and Campomelic dwarfism, but none of the other diagnostic anomalies of any such associated syndrome were present in our patient. Most common causes of death are aspiration penumonia, acute airway obstruction, starvation and right-sided heart failure. Micrognathia, short floor of the mouth and glossoptosis play a major role in producing the above mentioned complications[2,9]. The prognosis is exceptionally good provided the baby is managed properly during infancy. Postnatally there is a rapid downward and forward growth of the mandible associated with descent of the hyoid bone and elongation of the cervical cloumn. This leads to increase in size of the oral cavity relative to the tongue, and thus feeding and respiratory difficulties are diminished. The obstruction is almost completely relieved by the age of six months and a normal profile is reached by the age of three years., Function of organs become normal but some cosmetic deformity of the chin may remain, which is treated by plastic surgery.

The baby should be nursed in a prone or partially prone or position. Feeding needs great care and patience on the part of the nursing staff. Tube feeding may sometimes be necssary. Tongue suturing (glossopaxy) with the lower lip, to prevent glossoptosis till the mandible is sufficiently grown, is sometimes helpful.

The use of various splints and traction devices to pull the mandible forward have been successful and tracheostomy is usually not necessary.

Repair of the palate is usually under-taken at about the age of 12-18 months and later on dental referral is mandatory.

Conclusion

A potentially curable congenital anomaly like Pierre Robbins Syndrome, which needs delicate care and attentive management, is in fact a challenging job for the neonatologist. Education of the parents, excellent nursery care and regular follow-up remain the mainstays to ensure a completely normal life later on.

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