CASE REPORT

Apert Syndrome

by

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Abstract

A case of Apert syndrome in a male child of 5 months old has been reported. The diagnosis was based on the clinical appearance (phenotype) showing acrocephaly and syndactily of both hands and feet, supported by skull rontgenography and ultrasonography.

The patient was the third child from normal parents, and the two other children were normal. Apert syndrome is a genetic dominant automal disease; and because there were no other sufferer from the family history, the occurrence of this syndrome has been caused by a new mutation. Symptomatic therapy such as the administration of acetazolamide for hydrocephalus and vitamin suplement to improve his general condition, and even physical physiotherapy have been carried out. Genetic counselling to the couple has been provided as well.

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Introduction

Apert syndrome (acrocephalosyndactily) is a congenital multiple malformations. According to Durham, the term "syndrome" has been used in 1541 in Galen's book, and "syndrome" was defined as fixed abnormal pattern, of symptoms and signs at the same time [1].

Apert syndrome is a genetic disease inherited as dominant autosomal, and its incidence is 1 in 160.000 of births [2]. The phenotype of this syndrome is characterized by synostosis of the sutures of skull and face, for example in orbital synostosis will show exophthalmus, hypertelorism and strabismus. The syndactily of the hands and feet will cause severe psychologic depression to the patient and his parents. Other associated malformations in Apert syndrome may be congenital heart disease, central nervous system abnormality and mental disorder.

The management of Apert syndrome consists of conservative therapy and operative procedure, although the result was usually not rewarding.

In this paper a case of Apert syndrome has been reported to make the medical personels aware of the possibility of a certain syndrome in children showing multiple congenital malformations.

Case

A five month old male child was referred from Cilacap Regency Hospital to Dr. Sardjito General Hospital Yogyakarta with syndactily of hands and feet as well as a large head.

From alloanamnesis it was noted that defecation and mixtion were normal, the appetite was normal, afebrile, without vomiting nor respiratory tract infection and convulsion. The quality and quantity of nutrition were good. There were mental and physical growth retardation based on history. The mother has never used either drug or traditional medicine. She has never been sick during pregnancy, and she has been checked up regularly at maternal and child health clinic. The history of delivery revealed a spontaneous birth, full term, no asphyxia, assisted by a midwife, and his birth weight was 3300 grams.

Family history: his father age was 35 years old and his mother was 33 years of age. The patient was the third child of three children in this family. There was no other case of the same abnormality in the family either from the father side or from the mother side.

On physical examination the child showed a good general condition. He was councious; the pulse rate was 120/minutes regular; the respiratory rate was 24/minutes and axillary temperature was 36,5° C. The blood pressure was 90/60 mmHg. His nutritional status was 85% of standard Harvard P50 (the body weight of 6,5 kg).

The result of examination of this malformation syndrome (phenotype) were as follows. The head showed acrocephaly as a clover and the head circumference was 44 cm. He showed a wide forehead, flat face, depressed nose and wide fontanel. The orbital examination showed exophthalmus and hypertelorism. Deep arch of palatum and uvula bifida were found.

There were normal peripheral blood findings as well as stool and urine. The ultrasonographic examination of the skull showed a slight widening of the right and left ventricles with suspected hydrocephalus. Skull rontgenography revealed an abnormal head circumference. Rontgenography of the feet showed complete number of bones, but hands rontgenography showed overlapped position of phalanges, difficult to evaluate. Latex aglutination test for toxoplasmosis was negative.

DDST Denver Development Screening Test) was abnormal; it showed especially delayed on fine and gross motor. Otolaryngologic examination showed normal tympanic membrane, and the hearing reaction to child audiometry was positive. Anterior rhinoscopy showed normal findings.

A diagnosis of Apert syndrome was made. Symtomatic treatment given consisted of was: acetasolamide (Diamox) with the dose of 5 to 10 mg/kg body weight, and neutrophic vitamin supplement (Nootrophil) of 150 mg. Genetic counselling to the parents has been given as well.

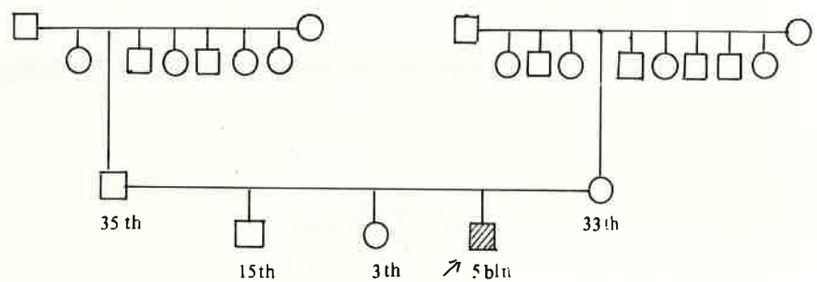

Figure 1 : *The family pedigree of the patient with Apert syndrome*

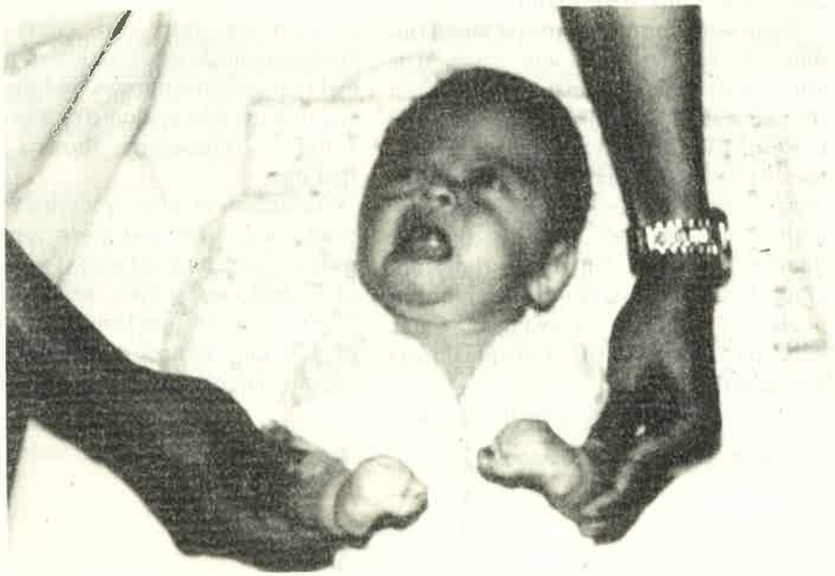

Figure 2 : *A photograph of the patient showing the syndactily of the hands.*

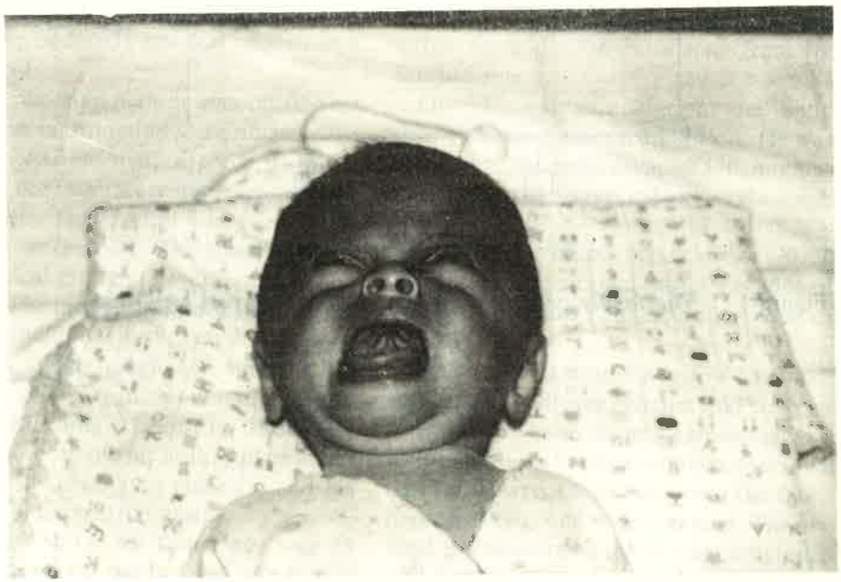

Figure 3 : *A photograph of t' 't showing a deep arch of the palatum*

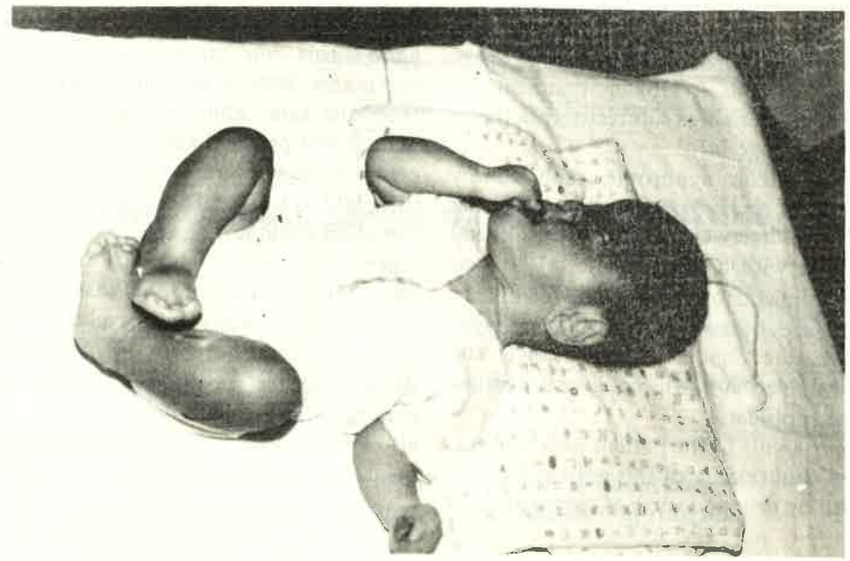

Figure 4 : *The clinical appearance (phenotype) of the patient suffering from Apert syndrome.*

Discussion

Apert syndrome was first reported by Wheaton in 1894. In 1906 Apert reported nine additional case, and until 1960 Black have recorded 150 cases [3]. The disease was a genetic disease inherited as autosomal dominant and most of them were caused by new mutation's. Apert syndrome is characterized by irregular craniosynostosis, mid facial hipoplasy and syndactily [3]. Its clinical figure is very characteristic, and this syndrome was one of craniosynostosis, i.e.: early sutures aposition of skull bone. There are three syndromes that belong to craniosynostosis : Crouzon syndrome (acrocephaly), Apert syndrome (acrocephalosyndactily) and Carpenter syndrome (acrocephalopoly syndactily). Three syndromes have a same phenotype i.e.: the abnormalities of head and face.

Protrusion of the eye balls in Crouzon syndrome is very severe, sometime it is followed by destruction of the eye balls. Raniwati et al., (1966) have reported one case of Crouzon syndrome [4]. To this case enucleation of one eye ball and tarsorraphy (the closure of the lids) of the other because of the eye ball destruction have been done. In Apert and Carpenter syndromes there were slight protrusion of the eye balls, but both also show syndactily, and Carpenter syndrome also shows polysyndactily (polydactily and syndactily).

In this reported case the clinical signs (phenotype) were very characteristic so it was easy to make a diagnosis clinically. Some additional examinations were not

done for supporting the diagnosis, but for predicting the severity of the disease and to find other possible malformations. According to Gills & Hogan treatment for craniosynostosis was suturectomy before 5 months of age [4].

In Crouzon syndrome reported by Raniwati et al., the suturectomy was done in the Neurosurgery Unit DR. Sardjito General Hospital with a good result [4]. It was very pesimistic for physical and mental abnormalities because those of the three syndroms were very severe, so medical and surgical procedure will not give a solution to the problems.

The parents of the patient were normal, so the abnormality of this patient must be caused by new mutation of spermatozoon [5]. If the couple want to have more child, there was no risk of recurrence. In the other hand if this child can live until adulthood and marry, 50% of the children will suffer from the same abnormality. Because of mental and physical abnormalities of this case were very severe, it was impossible for this child to live until adulthood, it means this child could not be able to transmit his Apert gene (natural selection). It is always important to follow up this case and also to do physiotherapy and psychotherapy.

Surgical procedure for the fingers of hands and feet is difficult because the abnormalities were not only syndactily, but also abnormalities of growth and development.

REFERENCES

1. HARTONO & SARODJA R. Sindrom penyakit genetik yang menyangkut kelainan mata. B I Ked 1987; 14 (2): 67-72.
2. FEINGOLD M, PASHAYAN H. Genetic's and birth defects in clinical practice. 1st ed; Boston : Little Brown, 1983;
3. YONES KL. Smith's recognisable pattern of human malformation, 4th ed, Philadelphia: WB Saunders, 1988.
4. RANIWATI, HARTONO, GUNAWAN. Penyakit Crouzon. KPPIM IV Perdami, Padang, 1986.
5. EMERY. Element of medical genetic. 4th ed; Edinburgh: Churchill Livingstone, 1975.