Case Report

Apert syndrome: a case report

Masoud Gharib
University of Social Welfare and Rehabilitation Sciences, Pediatric Neurorehabilitation Research Center, Tehran, Iran

Nazila Akbar Fahimi[1]
University of Social Welfare and Rehabilitation Sciences, Tehran, Iran

Apert syndrome is a genetic disorder first described by Eugene Apert in 1906. Its incidence is approximately one in 50000 births. This syndrome is associated with many abnormalities in the body and affects the central nervous system. Rehabilitation can increase children and their parent's quality of life. We report a case of Apert syndrome and the occupational therapy program conducted. In this paper We report a case of Apert syndrome and the occupational therapy program conducted. He was diagnosed by a neurologist upon physical examination.

Keywords: Apert syndrome, Rehabilitation

Submitted: 18 Dec. 2011
Accepted: 5 April 2012

Introduction

Over the past decades advances have been made in the prevention and treatment of developmental malformations. This revolution in our conceptualization of developmental anomalies has led to an improved ability to handle and prevent them. Despite these improvements, some conditions remain or continue to exhibit a large number of sporadic occurrences (1).

Apert syndrome is one of these conditions. It is a genetic disorder first described by Eugene Apert in 1906. Apert syndrome is known to be inherited as an autosomal dominant complex multisystem disorder, but most cases are sporadic. Although Apert syndrome is rare, its incidence is approximately one in 50000 births (4.5% of all craniosynostosis cases have Apert syndrome) (1, 2).

This syndrome is characterized by: craniofacial dysmorphy, hands and feet syndactyly and craniosynostosis (premature fusion of cranial sutures). Other anomalies include: congenital heart defects, central nervous system (CNS) abnormalities (including defect of corpus callosum and ventriculomegaly), limitation of shoulder movement, short humerus, urogenital anomalies, dermatologic manifestations, hearing loss, ear infection, sleep apnea, severe acne, poor intellectual development, and increased incidence of eye injuries (3, 4).

Apert syndrome has no known cure but orthopedic surgery can help correct the abnormalities. Surgery takes place in three steps: release of fusion, midface advancement and correction of wide set eyes. Other surgeries may be necessary depending on the individual's problems (such as: myringotomy, tracheotomy). Rehabilitation too can promote children and their parents' quality of life (5). We report a case of Apert syndrome and the occupational therapy program conducted. He was diagnosed by a neurologist upon physical examination.

Case report

"B.H." was an 11 year old boy. He has many aspects of Apert syndrome but no cardiac disorder. This young man was referred to the Occupational therapy clinic/ward because of psychomotor retardation at the age of 15 months. He is the first child of non-relative young parents with a history of epilepsy in the maternal aunts.

History during Pregnancy: Severe edema with hypertension since 5th month and amniotic leakage from 6th month.

History during Delivery: cesarean section (C/S) by 32 weeks due to rupture of membranes and hemorrhage. Birth weight was 2200 gm. He was admitted due to poor feeding and severe irritability by the 7th day.

Immunization: Up-to-date. He had high fever for 3 days after his first immunization.

1- All Correspondences to: Nazila Akbar Fahimi, PhD. Candidate in Occupational Therapy: Email: <fahimi1970@yahoo.com>

EEG: abnormal (slow background with scattered slow spike waves).
MRI: prominent brain sulci and ventricular system were noted for his age, indicative of brain atrophy. Corpus callosum was hypoplastic especially in the splenium part.
Abdominal ultrasound: normal
TORCH: normal
ABR: Bilateral sensorineural deafness
Ophthalmologic consultation: bilateral cataract (had been operated by age 2 months)
Chromosomal study: NL (Fig1)

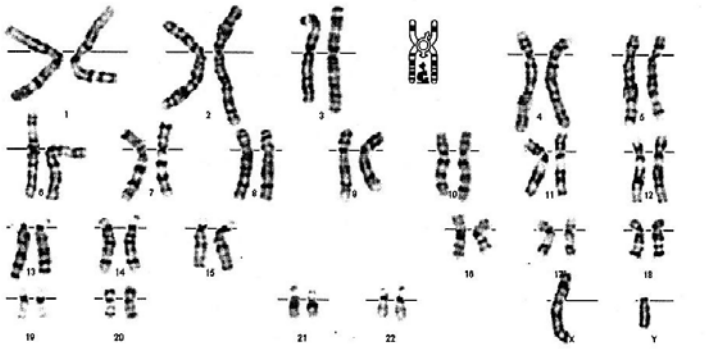

Figure 1: Twenty metaphase spreads were studied on the basis of GTG technique at 500 band resolution, revealing 46 chromosomes. No chromosomal aberration detected.

Metabolic studies including serum and urine analysis, serum NH3, lactate, Urinary reducing substances were normal.
Medication: Primidon 20 mg/kg/day
Growth & Development at first OT observation (by age 15 months): Head control on porn position, no sitting, no speech, no hand regard or using. He was irritable, hypersensitive, mild hip abduction restriction, large hand thumb, polydactylia of hands and feet; double and fused foot thumb. He made poor eye contact, had no organomegaly or skin disorder, and had trouble swallowing.
He had surgical operation for polydactylia by 19 months.

Occupational therapy interventions:
OT programs for B.H. was based on Neurodevelopment facilitation techniques, feeding program and dark room. After 12 months OT intervention and 47 sessions, he was more cooperative, more alert and had fair swallowing capability. He made no tremendous motor system improvement, however he could control his head, sit with support, roll to both sides and react to others.
OT programs for his parents included: 1) Activity Daily Living (ADL) training such as bathing, bed mobility, dressing and eating. 2) Injury prevention or reduction, education and safety awareness training during his care with proper methods and suitable devices. 3) Emotional and social support to cope with this condition and 4) teaching coping strategies. After 12 months of the OT program his parents had learnt caring methods very well and their quality of life had improved significantly (according to the SF-36 questionnaire).

Conclusions

Apert syndrome is rare and has no known cure. Parents of children affected may lack knowledge and skills in specific areas of parenting, such as mobility, providing for a child's safety, feeding, or daily routine care. The rehabilitation team especially occupational therapists can provide these services. They can help parents cope with their condition and to participate fully as their role of parents.

Refrances:

1. Tolarova MM, Harris JA, Ordway DE, Vargervik K. Birth prevalence, mutation rate, sex ratio, parents' age, and ethnicity in Apert syndrome. American Journal of Medical Genetics. 1997; 72 (4) : 394-8.
2. Freiman A, Tessler O, Barankin B. Apert syndrome. International Journal of Dermatology. 2006; 45 (11) : 1341-3.
3. Cohen MM, Kreiborg S. The central nervous system in the Apert syndrome. American Journal of Medical Genetics. 1990; 35 (1) : 36-45.
4. Faro C, Chaoui R, Wegrzyn P, Levaillant JM, Benoit B, Nicolaides KH. Metopic suture in fetuses with Apert syndrome at 22-27 weeks of gestation. Ultrasound in Obstetrics and Gynecology. 2006; 27 (1) : 28-33.
5. Glaser RL, Broman KW, Schulman RL, Eskenazi B, Wyrobek AJ, Jabs EW. The Paternal-Age Effect in Apert Syndrome Is Due, in Part, to the Increased Frequency of Mutations in Sperm. The American Journal of Human Genetics. 2003; 73 (4) : 939-47.