TRISOMY 18 in a 50-year-old female

B. Bhanumathi, Neelam A. Goyal, Z. A. Mishra

Smt. Motibai Thackersey Institute of Research in the Field of Mental Retardation run by "The Research Society for the Care, Treatment and Training of children In Need of Special Care" Sewri Hill, Sewri Road, Mumbai - 400 033, India

She was brought to our Institute at the age of 31, with speech delay and mental handicap. She was assessed by the multidisciplinary team in the institute to determine the cause of her problems. Clinical evaluation revealed dysmorphic facial features, microbrachycephaly, camptodactyly, clinodactyly, abnormal dermatoglyphics and severe mental handicap. Cardiovascular system examination was normal. Chromosomal analysis revealed a trisomy of Chromosome 18. The phenotype of trisomy 18 and the rarity of prolonged survival in this case are discussed.

Key words: Mental retardation, Edward syndrome, life span

Introduction

Trisomy 18 or Edward’s Syndrome is a specific and well-documented syndrome like trisomy 21 or Down syndrome, but is much rarer. It was first described by Edward et al in 1960.[1] It is the second most common multiple malformation syndromes, with an incidence of about 0.3 per 1000 newborns with a preponderance of males to females.[2] The majority of cases have full 18 trisomy as a result of nondisjunction and rarely due to translocation as in trisomy 21.[3] The result of faulty chromosomal distribution is most likely to occur at older maternal age - the mean maternal age of babies born with this syndrome being 32 years.[3] A bimodal distribution with two peaks at 25-30 years and 40-45 years is described.[4] The phenotype is characterized by a severe malformation syndrome, almost always leading to death in the early weeks of life.[5] The mean survival is two to three months for males and 10 months for females.[4] Few patients have survived to the age of 15 or 19 years.[6,7] Patients with mosaicism have prolonged survival rate.

Case Report

She was brought to our institute at the age of thirty one for investigation and guidance, with the chief complaints of mental sub normality and limited speech. She was the only child born to nonconsanguineous, Hindu parents. Her father and mother were 30 and 27 years old respectively at her birth. She was born at term and birth weight was 2.250 kg. She did not cry immediately after birth and was given physical stimulation. She also had sucking difficulty and aspirated on the third day of life and was given nasal oxygen. The antenatal period was reportedly uneventful.

Motor and mental development was slow from birth. Feeding and chewing difficulties persisted till the age of eight years. At six months of age, she had high fever, for which she was treated with antibiotics. She underwent tonsillectomy at eight years of age. Onset of menarche was at 18 years. Hysterectomy was performed at 28 years of age. There was no history of convulsion or neuroregression. Other systemic examination was normal.

Family history revealed mental deficiency in her paternal great grandmother and paternal grand aunt’s daughter while her father’s cousin was mentally ill.

On clinical examination, her height was found to be 154 cms. And she weighed 44 kgs. Microbrachycephaly, bushy eyebrows, synphorisis, ocular hypotelorism, concomitant divergent squint, antimongoloid slant of eyes, beaked nose, low set ears, high arched palate, dental malocclusion, left simian crease, right clinodactyly, camptodactyly of the fifth finger and both index fingers and hypoplastic nails in both fifth fingers were present.

Address for Correspondence: B. Bhanumathi, Smt. Motibai Thackersey Institute of Research in the Field of Mental Retardation run by "The Research Society for the Care, Treatment and Training of children In Need of Special Care" Sewri Hill, Sewri Road, Mumbai - 400 033, India. E-mail: bahanatu@gmail.com

Indian Journal of Human Genetics September-December 2006 Volume 12 Issue 3
Psychometric testing revealed an I.Q. of 23 to 25 points, which placed her in the severe grade of mental retardation. Hearing and visual acuity were within normal limits. Her speech was limited to simple sentences and was dyslalic and slurred. Hypertonia was also present.

Routine investigations were normal. Plasma and urinary amino acid chromatogram showed normal pattern. Roentgenogram of the chest, 2-D Echocardiography and abdominal ultrasonography were normal. X-rays of the hip joints revealed osteochondritis of epiphyses of the roof of both acetabulae. Chromosomal analysis of over 100 metaphases revealed a free trisomy of Chromosome 18.

Discussion

Patients of trisomy 18 generally present a clinical picture that is readily recognizable. There is however a considerable degree of overlap with trisomy 13 or Patau syndrome.[8]

The most common clinical features are: polyhydramnios, small placenta, single umbilical artery, mental retardation, weak cry, hyper tonicity, feeding difficulty, failure to thrive, flexion of fingers with the index overlapping the third finger and the fifth overlapping the fourth, cryptorchidism, dorsiﬁcation of halluces, interventricular septal defect, patent ductus arteriosus, inguinal and/or umbilical hernia and short sternum. Less commonly seen are epicanthus, tallips calcaneovalgus, “rocker-bottom” feet, soft tissue syndactyly of the second and third toes, Meckel’s diverticulum, heterotrophic pancreatic tissue and thin diaphragm with evagination.[9]

The most common oral and facial anomalies include prominent occiput, narrow bi-frontal diameter and mild hirsutism of forehead, low-set malformed ears and micrognathia. Less commonly noted features are microcephaly, corneal opacity and ptosis of eyelids. Cleft lip and cleft palate have also been noted.[3]

In our case, the typical facial dysmorphic features were absent though failure to thrive, feeding difficulty, mental deﬁciency, microcephaly, hypertonicity, ﬂexion deformities of ﬁngers were present. Cardiovascular abnormalities were absent.

Survival in trisomy 18 is related to the severity of congenital malformations and to some extent, the availability of paediatric care. Survival into childhood or beyond has been noted on rare occasions; when they have lived to the age of 15 or 19 years.[6,7] However, review of literature suggest that our patient has the longest survival which could be due to the absence of cardiovascular anomaly. More than 100 metaphases were screened to rule out mosaicism as it has been reported that patients with mosaicism have prolonged survival.

Acknowledgments

We are grateful to Mrs. T. D. Shroff, our Honorary Administrative Director and the board members of the “Research Society” for their kind permission to publish the above data. Our thanks are also due, to all our colleagues for their support and suggestions.

References


Source of Support: Nil, Conflict of Interest: None declared.