LETTER TO EDITOR (VIEWERS CHOICE)

CONGENITAL TALIPES EQUINES VARUS AS AN ADDITIONAL MALFORMATION OF BARDET-BIEDL SYNDROME

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A 2-year-old boy from Tiruvanamalai (Tamil Nadu, India), the 3rd child of consanguineous married couple, born after full term gestation by normal vaginal delivery at home without birth asphyxia, presented to us with difficulty in walking, deformity of left foot and delay in speech. There was no history of convulsions. As per parents, vision and hearing was normal. He was on cow's milk with bottle till the age of 9 months. Presently he is receiving around 800-900 cal/day. On examination the delay in developmental mile stones, polydactyly of the left hand and left sided congenital talipes equino varus (CTEV) were detected. Anthropometric measurements revealed infantile obesity (weight-15 kg , length-74 cm, upper segment: lower segment ratio of 1.4, BMI of 27.7%, waist circumference -57 cm, head circumference - 40 cms, chest circumference-60.5cm with waist circumference: head circumference of 1.05). The examination of cardio-vascular system and genitalia revealed nothing abnormal. Apart from these findings nothing positive was detected clinically. His fundus examination revealed features of retinitis pigmentosa. BERA and echocardiography were normal. Ultrasound abdomen revealed normal kidneys. IQ was 70. Karyotype of the patient was normal - 46 XY, but 28% of the metaphase showed different chromosomal aberrations like breaks, gaps, fragments and isochromosomes. The 50 % of the metaphase showed acrocentric associations (satellite associations) indicating the presence of DNA damage. Because of the constraints we were not able to mark the locus. The child thus was detected to have Bardet-Biedl syndrome with talipes equino varus malformation.

Bardet-Biedl syndrome (BBS) is rare genetically heterogeneous autosomal recessive disorder with incidence of less than 1:160000 (1). The syndrome is characterized by progressive retinal dystrophy, polydactyly, obesity, hypogonadism and mental retardation. In some cases, literature shows the cause of death is renal failure (1, 2). It is now widely accepted that mutated BBS genes affect normal cilia functions, which in turns, cause BBS. The gene products encoded by these BBS genes, called BBS proteins, are located in the basal body of the cilia of the cell. Currently 12 gene mutations causing BBS have been identified. (3) BBS is a ciliopathic disorder. Other known ciliopathies include primary ciliary dyskinesia, polycystic kidney and liver disease, nephronophthisis, Alstrom syndrome, Meckel-Gruber syndrome and some forms of retinal degeneration(4). Its affection is global. A variety of malformation involving different body systems has been identified. Here we report a case of BBS with an additional malformation of CTEV. The pigmentary retinopathy, poor visual activity, and/or blindness in eyes (5), anosmia in nose, polydactyly or syndactyly in hand and foot, fibrosis, situs inversus and Hrischsprung disease in gastrointestinal system, hypertrophy of interventricular septum and left ventricle, and dilated cardiomyopathy in cardiovascular system, hypogonadism, renal failure, uterus duplex, septate vagina and hypoplasia of uterus, ovaries and fallopian tubes in urogenital system have been identified (1, 2, 4). The mental and growth retardation, behavior and performance deviations including subnormal social interactions in the shape of mild-autism and defective thermo-sensations have also been reported (4).

Apart from the cardinal features viz. infantile obesity, retinitis pigmentosa, polydactyly, mental retardation, the talipes equino varus malformation was also noted in our case. The CTEV malformation was also detected in the child's 7 months old younger brother. His focusing towards light was absent which indicated impaired visual activity. His social interactions were also at the minimum level and hearing was defective. Till date the involvement of skeletal system has been reported in the shape of postaxial polydactyly, syndactyly, brachydactyly and clinodactyly (6). But in our encounter with this BBS South Indian family both brothers were suffering from talipes equino varus malformation, which is a neoclinical manifestation of the involvement of skeletal system.

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