Brief Reports

Body Stalk Anomaly

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The term body stalk anomaly is given to a condition with a large irregular abdominal wall defect associated with multiple abnormalities and a very short umbilical cord(1). We are reporting three such cases registered in our department.

Case Reports

The three reported still births were referred to us for evaluation of cogenital anomalies. The striking features in all the three cases were gross spinal deformity with shortening, irregular abdominal wall defect with protrusion of entire liver and bowel, covered by a thin sac which was connected to the placenta by a shot umbilical cord (*Fig. 1*). All were born to couples with poor reproductive history. There was no history of exposure to teratogens of either of the couple during peri-conceptional period or of the mother during the antenatal period in any case. In the first case diagnosis was made by ultrasonography by the presence of a very short umbilical cord connecting the placenta to a sac through which liver and bowel were protruding through the abdomen. The salient features of all the three cases are given in *Table I*.

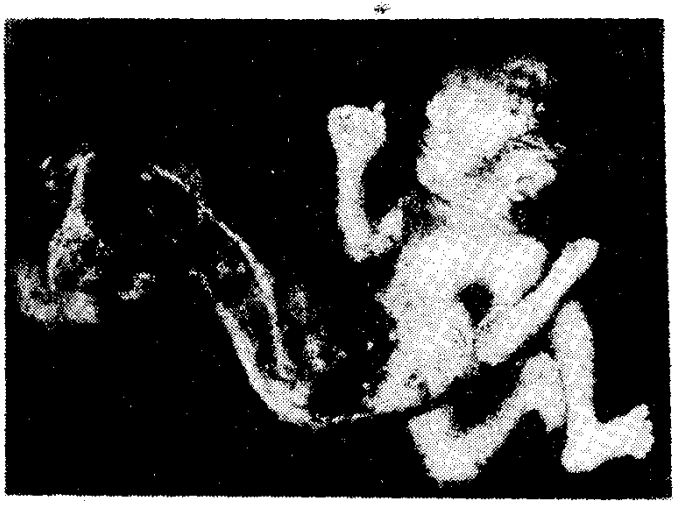

Fig. Photograph of Case 2 showing short cord and placentsa: fetus with liver and intestine protruding.

Discussion

Table II gives the features of various types of abdominal wall defects. In body stalk anomaly, the defect in the anterior abdominal well is large and irregular. Most of the abdominal organs protrude out and are covered by a thin sac. A short umbilical cord is attached to the apex of the sac. They are almost invariably associated with major spinal and pelvic deformities, hypoplastic lower limbs and closed neural tube defects (NTD) among others(2). This defect is very rare. Incidence is quoted as 1 in 50,000 occurring predominantly in males(3)

Abdominal muscle deficiency anomaly

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TABLE I – *Summary of Clinical Features*

Feature	Case 1	Case 2	Case 3
Maternal age (yrs)	29	24	27
Paternal age (yrs)	34	34	36
Consanguinity	No	III degree	II degree
Reproductive history	Infertility for 10 yrs Treated with clomiphans	1 abortion at at 5 months	4 abortions
Live children	Nil	Nil	Nil
Prenatal	Diagnosed by US as body stalk anomaly after 20 weeks	Diagnosed by US as IUGR and omphaloceala after 20 weeks	Not done IUD
Delivery	Induced with bougie; aborted	Induced with bougie; aborted	Induced-failed; bougie hysterotomy
Placenta	Normal	Normal	Macerated
Umbilical cord	Very short, 15 cm	Very short, 20 cm	Very short 15 cm
Weight for gestation	SFD	SFD	SFD
Abdominal sac	Umbilical cord attached to apex of the sac	Cord attached to apex of the sac	Not made out
Contents of sac	Liver, spleen intestine	Liver, intestine	Not made out macerated intestine
Neck	Very short	Very short	-
Thorax	Short Kyphosis +	Short Kyphosis +	Short
Limbs	Small limbs bilateral CTEV	Small asymmetrical lower limbs Bilateral CTEV	Small
Genetalia	Small phallus No scrotal sac	Small phallus Small scrotal sac	
Anal orifice	Absent	Absent	
Autopsy	Heart pushed down; testes intra abdominal; Kidney-small cysts	Heart pushed down; testes intra-abdominal; kidneys small	Macertated not done
Karyotyping of fetus from cord blood	Not successful	46 XY	Not done
Father	46 XY	Not done	46 XY
Mother	46 XX/47 XXX Mosaic	Not done	46 XX

TABLE II—*Anterior Abdominal Wall Defects*

Features	Gastroschisis	Exomphalos	Bod stalk defects
Size	Small	Varies Minor, Major	Large
Edges	Smooth	Smooth	Irregular
Site	Close to normal cord	Involving umbilical cord	Involves umbilical cord
Covering sac	No	Translucent Sac wall	Filmy sac
Umbilical cord	Normal	Defect involves Cord	Short
Protruding organs	Loops of bowel	Minor-Herniation of intestine into cord Major-bowel and liver	Almost whole liver and bowel
Associated defects	10%-atretric intestine, Congenital heart disease	50% neural tube defects, Congenital heart disease	100%-spinal and pelvic deformity, closed NTD, undescended ex-atrophy, hypoplastic lower limbs
Karyotype	Normal	Abnormal in 30% (trisomies and others)	Normal

consisting of abdominal wall hypoplasia, urinary tract dysplasias and cryptorchidism among other defects may be the consequence of a single localized defect in early mesoderm which eventually contribute to the formation of abdominal musculature, urinary tract musculature, renal parenchyma and possibly gubernaculum testes. Defective musculature can effect fetal movements and this fetal akinesia may be responsible for the very short umbilical cord as umbilical cord growth depends on tensile forces of fetal movements(4). No chromosomal abnormality has been reported with this defect. No specific genetic or environmental cause has been implicated and recurrence has not been reported.

The reproductive history is significant in our cases with relative infertility and/or habitual abortion. There are no live children in any of the 3 families reported. However, one mother had a triple-X mosaic karyotype. Abnormal meiotic division occurs in triple-X states. Such women are known to be infertile due to oocyte stresia. They may have chromosomally normal offspring, or offspring with anomalies without chromosomal aberration(5).

This condition can be diagnosed prenatally by ultrosonography.

Acknowledgement

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WAGR Complex

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Various epidemiological studies have shown aniridia to be increasingly associated with Wilms' tumor(1,2). The association of sporadic Aniridia with Genitourinary anomalies and mental retardation particularly makes a child high risk for the development of Wilms' tumor. Association of these four entities have been given the acronym WAGR complex. Only 29 patients of this complex were described till 1986(3). A few more have been added since then(4). We report a case of WAGR complex with Stage V Wilms' tumor treated with multimodal therapy.

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Case Report

A 2½-year-old boy was admitted to the Department of Pediatric Surgery, SPMCHI, Jaipur with complaints of asymptomatic mass abdomen for three months duration. There was no history of weight loss, hematuria, *etc.* Examination revealed anemia, bilateral hypoplasia of iris, obvious cranio-facial dysmorphism with abnormally large pinnae (*Fig. 1*). Nys- severe photophobia was noted. The child was severely mentally retarded. Abdominal examination revealed 10 × 8 cm firm retroperitoneal mass in the right lumbar region. The left kidney was also palpable. Examination of genitalia revealed severe penoscrotal hypospadias with right impalpable testis. A clinical diagnosis of WAGR complex was made with a likelihood of

Fig. 1. Photograph of the child. Note the large pinnae and obvious cranio-facial dysmorphism. The child has closed his eyes due to severe photophobia.