MULTIPLE SEVERE CONGENITAL MALFORMATIONS OF THE FOETUS—A CASE REPORT

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Congenital anomalies of the foetus as observed from different reports have ranged from 2 to 6.5 per 1000 livebirths, the incidence being higher among the stillbirths. However, the combinations of abnormalities and involving many systems, is very rare (Rubin, 1967). Multiple anomalies constitute only 11% of all malformations (Anderson and Reed 1954). The occurrence of multiple congenital abnormalities in a foetus of indeterminate sex with absence of anal opening, genito-urinary abnormalities, omphalocoele, lordosis and scoliosis of the spine, thoracic cage deformity prompted us to report the following case.

CASE REPORT

R. D. 30 years, 5th gravida, P4 + 0 was admitted on 13–2–78 at 4 a.m. with 38 weeks' amenorrhoea. She had hydramnios during this pregnancy. She started labour pains and ruptured membranes 10 hours before admission. She had a bout of vaginal bleeding just before admission. She denied any history of vomiting, drug intake or exposure to radiation during this pregnancy. She had pyrexia for 3 days, not associated with any other symptoms at the 8th week of pregnancy and she took no medication for it.

She had 4 full term normal deliveries at home. Her first pregnancy was associated with hydramnios and at term resulted in the birth of an anencephalic male fresh stillbirth. The other 3 pregnancies were normal and all the 3 children were alive and healthy.

There was nothing significant in the past, family or menstrual history. No history of congenital defects in the relatives.

On examination, patient looked pale. Pulse 96/minute, regular. Blood pressure 110/80 mm Hg. Abdominal findings were consistent with 32 weeks pregnancy with breech presentation. Foetal heart sounds were not heard. Vaginal examination excluded placenta previa. Patient was given syntocinon drip and had breech delivery at 11 a.m. the same day. She had no postpartum haemorrhage and received one unit of blood. The foetus was a fresh stillbirth. The foetus and placenta weighed 2 Kgm. Placenta was large and could not be separated off from foetus as the amnion was adherent to the anterior abdominal wall. Foetus showed multiple severe malformations.

1. The thoracic cage showed deformity. The right side of thoracic cage was well developed and sternum was deviated to the left (Fig. 1). Left scapula was better developed compared to the right. There was gross abnormality of the spine—scoliosis with convexity to the left. There was marked lordosis of the lumbosacral spine (Fig. 2). The lower limbs manifested talipes equinovarum deformity (Fig. 4).

2. The anterior abdominal was absent and there was a huge abdominal hernia comprising of liver, spleen gut (Fig. 3). The attachment of umblical cord was situated beside the margin of the hernia.

Umblical cord was poorly developed and represented by a thin 12 cm long streak containing a single vessel attached to the amnion, which was forming the sac of the hernia.

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3. External genitalia consisted of two labioscrotal swellings, beneath and in between these there was a solitary opening; on probing that led to opening in the anterior abdominal wall (Fig. 4). There was no bladder, uterus or adnexae found in the pelvic cavity. There was no anal opening.

Investigations done viz. Blood Grouping, blood sugar curve, serological tests for syphilis, tests for toxoplasmosis were normal.

Comment

Major abnormalities form 9.2 per cent of all malformations (Pedlow, 1961). Earlier, Quingley (1935) found the incidence to be 10%. The combination of congenital anomalies involving genitourinary, skeletal system, ohphalocele with indeterminate sex and absence of anal opening reported in the present case is very rare. Peterson *et al* (1970) reported in a male foetus born to a diabetic mother treated with insulin and dibein (phenforminichlorid, an oral hypoglycemic agent) somewhat similar abnormalities namely, pronounced scoliosis of the spine, huge abdominal and thoracic hernia, moderate micrognathia and micromelia left arm with agenesis of radius and metacarpal bones.

Lal (1973) reported a male stillborn monoamniotic twin with absence of external genitalia and anal opening, umblical hernia and abnormalities of lower limbs.

It is difficult to find out the etiology of such severe malformations in this case. It might have been due to some genetic defect in the fertilized ovum giving rise to multiple congenital abnormalities. The arrest of growth of some organs and asymmetrical growth of the rest might have been initiated in second or early third months. The patient had a febrile episode at that time. It is difficult to say if that was of viral nature so as to hamper the intrauterine growth. The problem of recurrence of the abnormalities in this case does not arise as the patient had abdominal tubectomy.

Summary

Multiple congenital anomalies in a foetus of indeterminate sex with absence of anal opening, genitourinary abnormalities, omphaloccele, lordosis and scoliosis of the spine, thoracic cage deformity and a rudimentary umblical cord with a single vessel is reported. The pregnancy was associated with hydramnios and patient had given birth to anencephalic still born foetus previously.

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See Figs. on Art Paper II