

Umbilical Cyst with Edward Syndrome

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ABSTRACT

The advances in Obstetric ultrasonography have allowed early and accurate detection of anomalies in utero. Umbilical cord anomalies may include cysts, vascular anomalies and masses. Cysts detected in first trimester are mostly transient and the children are born normal. If detected in second or third trimester,

the risk of fetal anomalies are high, and warrants a chromosomal analysis. We present a case report where antenatally cord cyst was detected in second trimester, with the fetus progressing to have oligohydramnios and Intrauterine growth retardation (IUGR). The neonate was born with Edward syndrome.

Keywords: Chromosomal anomalies, Karyotyping, Obstetric ultrasonography, Pseudocyst, Umbilical Cord anomalies

CASE REPORT

A second gravida mother, with a normal first pregnancy and no history of abortions, was on regular antenatal follow-up. First trimester was uneventful and ultrasonography was normal. The second trimester ultrasonography revealed an umbilical cyst of size 40×15 mm. No other abnormalities were detected and placenta was normal. Parents were counselled regarding the risk of fetal anomaly and advised amniocentesis for chromosomal analysis. But parents didn't agree and chose to continue the pregnancy. Case was closely monitored throughout the gestational period. The fetus had intrauterine growth retardation with oligohydramnios. Full term female child was born with birth weight of 1800gm.

The neonate delivered had dysmorphic features resembling Edward syndrome—prominent occiput, low set deformed ears, micrognathia, cleft palate, upper limb

flexion deformity with clenched fists and overlapping fingers, kyphoscoliosis, genu recurvatum, overlapping toes and rocker bottom feet [Table/Fig-1-3]. Postnatal 2D echocardiography was normal. Ultrasonography of the skull revealed bilateral lateral ventricle dilatation. Karyotyping confirmed the diagnosis of trisomy 18. The neonate was discharged after one week of uneventful stay in the Neonatal Intensive Care Unit. Parents were counselled regarding child care and prognosis. The child expired at home in neonatal period. This case is being reported because the combination by itself is a rare entity. Moreover, there is no such case report from India.

DISCUSSION

Ultrasonography is a routine investigation performed during antenatal period. Increasingly it is being used as a non-invasive modality to detect fetal anomalies,



[Table/Fig-1]: Clinical photo of patient with Edward syndrome. **[Table/Fig-2]:** Edward syndrome (Low set ears, retrognathia and overlapping fingers). **[Table/Fig-3]:** Edward syndrome (Rocker bottom foot and genu recurvatum).

in addition to the usual indications of monitoring fetal viability, gestation and growth. Umbilical cord develops at about 7th week of gestation, and usually can be seen in ultrasonography by around 8th week. The exact cause of umbilical cyst is not known, but it is thought to be due to raised hydrostatic pressure in the umbilical vessels. The cysts are classified as true cysts and pseudocysts. True cysts have an epithelial lining- cuboidal or flat epithelium, and are remnants of allantois or omphalomesenteric duct. They are usually found near the fetal end, and are associated with genitourinary defects such as patent urachus, omphalocele, or gastrointestinal defects. Pseudocysts on the other hand lack epithelial lining, and arise from focal oedema or degeneration of Wharton's jelly. They are found in association with omphalocele and chromosomal anomalies such as trisomies. The type of cyst can be confirmed by histopathological examination.

Usually the cysts detected in first trimester, especially if they are single, small and having smooth borders, and located in middle portion of the cord are transient, and healthy babies are born [1,2]. Invasive tests are not typically needed, only ultrasonography monitoring being sufficient.

Detection of cysts in second or third trimester, or persistence from first trimester, large size, location near fetal or placental end, are associated with higher risk of fetal anomalies, with varying degrees based on different studies [3-5], even as high as 50%. The aneuploidies known to be associated are trisomy 18, 13 and 21. Thus, in such cases chromosomal analysis may be warranted [6].

Edward syndrome or trisomy 18 is fetal aneuploidy characterized by addition of an extra copy of chromosome to chromosome 18. The incidence of trisomy 18 is 1 in 6000 live birth and is associated with high rate of intrauterine deaths. Dysmorphic features of Edward syndrome consist of intrauterine growth restriction, small cranium with prominent occiput, low set ears, micrognathia, cleft palate, clenched hands with overlapping digits, hypoplastic nails and rocker bottom foot. Other anomalies include congenital cardiac, renal

and brain malformations. One should highly suspect the disorder if antenatal ultrasonography is suggestive of growth retardation, oligohydramnios or polyhydramnios, limb deformities, cardiac and brain or neural tube anomalies. Prognosis of Edward syndrome is extremely poor with 90% of infant deaths within six months of age.

There are few case reports about the rare combination of umbilical cord cyst with trisomy 18, but no such reports from India [7,8].

CONCLUSION

Detailed ultrasonographic examination of fetus is recommended when umbilical cyst is detected antenatally, especially in second or third trimesters, and any associated defects should be carefully looked for. Karyotyping analysis should be done in suspicious cases. Risk-benefit assessment and detailed counseling including option for termination of pregnancy should be offered to such parents.

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