

IMAGES IN CLINICAL PRACTICE

UMBILICAL CORD SWELLING IN A NEWBORN WITH DOWN'S SYNDROME

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A male baby born at 40 weeks of gestation as a fourth live birth from the fourth pregnancy of a 40-year-old mother was admitted to our neonatal intensive care unit (NICU) with a phenotype of Down's syndrome and respiratory distress. The double marker test performed during the prenatal period showed a high probability of having Down's syndrome. Physical examination of the neonate revealed upward slanting eyelids, a flattened face, especially the bridge of the nose, low-set small ears, palmar crease, and poor muscle tone with birth weight of 2730 g (3-10th centile), height of 46 cm (25-50th centile), and head circumference of 35 cm (50-75th centile). Along with the normal size of the umbilical ring, the umbilical cord was 2x2 cm in size and had intestinal tissue in it (Figure 1). In addition, except for a 2/6 systolic-diastolic murmur especially at pulmonary area, other systemic findings were normal. Echocardiographic examination revealed atrial septal defect and patent ductus arteriosus. Chromosomal analysis revealed 47, XY, +21 karyotype. The child underwent surgery for the umbilical cord swelling.

What is the umbilical cord swelling?

Umbilical cord hernia (UCH). The most common fetal anterior abdominal wall defects are omphalocele and gastroschisis.¹ UCH is another anterior abdominal wall defect that is not well defined and often misdiagnosed as minor omphalocele. Unlike a minor omphalocele, which refers to a true abdominal wall defect, congenital UCH is the herniation of the small intestines and sometimes other viscera into the umbilical cord due to the unsuccessful closure of the umbilical ring or the failure of the physiologically herniated intestines to fully withdraw into the abdominal cavity.² Omphalocele and UCH are both formed by the protrusion of internal organs from the umbilicus, but they have different embryological origin, morphology and prognosis.³ Omphalocele is associated with congenital anomalies, and therefore its prognosis is poor.³ However, UCH is rare and similar to the present case, its prognosis is excellent as there are no associated abnormalities.³

A large part of the intestines begins to hernia physiologically at the seventh week of pregnancy, towards the proximal part of the umbilical cord, called the extracoelomic cavity.⁴ This physiological herniation is necessary for the proper rotation of the intestines and sufficient growth of the fetus to accommodate the expanding intestines.⁵ Between ten and twelve

Figure 1. Umbilical cord swelling which contains intestinal tissue.



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weeks of gestational age, the intestines return to the abdominal cavity, leaving behind the umbilical cord, the umbilical ring often closes, and the extracoelomic cavity disappears.⁵ In rare instances, the umbilical ring is unable to close, and the variable parts of the intestines remain in the extracoelomic cavity as a congenital hernia to the umbilical cord.⁴

Omphalocele often occurs in association with other birth defects or specific genetic syndromes, such as Down's syndrome.⁶ Best to our knowledge, there are no association between the Down's syndrome and UCH. Therefore, this instructive case of Down's syndrome with UCH was presented.

Author contribution

SB and MA designed the study; SB and ET wrote the manuscript; UB operated the patient; ET and MA cared of the patient. All authors read and approved the final manuscript.

Compliance with Ethical Standards

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