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Pancreatic hamartoma in a premature Trisomy 18 female

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ABSTRACT

Pancreatic hamartomas are extremely rare tumors in adults and even more so in children. They are lesions characterized by acinar, islet and ductal components found in varying proportions and in a disorganized pattern. We report a case of a premature female with trisomy 18 diagnosed by amniocentesis. The newborn was delivered by cesarean section at thirty-three weeks of gestation and expired within one hour of birth. Postmortem examination exhibited numerous features associated with Trisomy 18 including lanugo on the torso and arms, micrognathia, microstomia, left low-set ear with small flat pinna, closed ear canal, clenched fists with overlapping fingers, rocker-bottom feet, narrow pelvis, large right diaphragmatic hernia and left pulmonary hypoplasia. Microscopic examination of the pancreas revealed an area, 1.2 cm in greatest dimension, with branching ducts and cysts lined by cuboidal epithelium intermingled within primitive mesenchymal proliferation and exocrine glands. The cysts measured up to 0.2 cm and were surrounded by a collarette of proliferating spindle cells as highlighted by Masson's trichrome stain. A diagnosis of pancreatic hamartoma was rendered. A total of thirty-four cases of pancreatic hamartomas have been reported in the literature including twenty-seven in adults, five in children and two in newborns. Our case may be the third pancreatic hamartoma reported in association with Trisomy 18. We recommend that careful examination of the pancreas be performed in individuals with Trisomy 18 to further characterize this lesion as one of the possible abnormal findings associated with this syndrome.

Keywords

Edwards Syndrome, hamartoma, pancreas, pancreatic neoplasm, Trisomy 18

CASE REPORT

The premature female was born at 33 weeks of gestation to a 25 years old mother, gravida 3 para 2, with a history of pre-eclampsia in the previous pregnancy. In this pregnancy, the fetus was found to have Trisomy 18 karyotype by amniocentesis. Fetal ultrasound at 21 weeks gestational age showed left displacement of the heart and absent nasal bone. The mother presented to at 33 weeks of gestation for

premature rupture of membranes and contractions. Fetal ultrasound performed at that time showed normal amniotic fluid volume, intrauterine growth restriction with estimated fetal weight of 1519 grams (less than the 10th percentile for gestational age), right side congenital diaphragmatic hernia with liver herniating into the thorax, left mediastinal shift of the heart, hypoplastic left lung, and clenched hands with

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limited movement of upper extremities suggestive of arthrogryposis. Doppler interrogation of umbilical artery was normal. The baby was delivered by cesarean section.

At the time of birth, the infant was floppy and apneic for approximately 8 minutes requiring stimulation and intermittent positive pressure ventilation. APGAR score was 1, 2 and 4 at 1, 5 and 10 minutes, respectively. Intubation and resuscitation were not performed given the poor prognosis, and she expired at less than one hour of life.

On postmortem examination, the newborn exhibited numerous features associated with Trisomy 18 which included the following: lanugo on the torso and arms, micrognathia, microstomia, left low set ear with small flat pinna and closed ear canal, bilateral clenched fists with index and fifth fingers overlapping

the third and fourth fingers, rocker-bottom feet, narrow pelvis, large right diaphragmatic hernia, and left pulmonary hypoplasia. No cardiovascular and genitourinary defects were identified. The placenta was small for the gestational age weighing 250 grams (normal is 342 grams for 33 weeks of gestation).

Although no abnormalities were noted on gross examination of the pancreas, microscopic examination revealed an area measuring 1.2 cm in greatest dimension characterized by branching duct-like structures and cysts lined by cuboidal epithelium that measured up to 0.2 cm. Primitive mesenchymal proliferation and exocrine gland formation were found in between (Figure 1A, 1B and 1C). A proliferation of spindle cells forming a collarette around the cysts were highlighted by Masson's trichrome stain (Figure 1D). A diagnosis of pancreatic hamartoma was rendered.

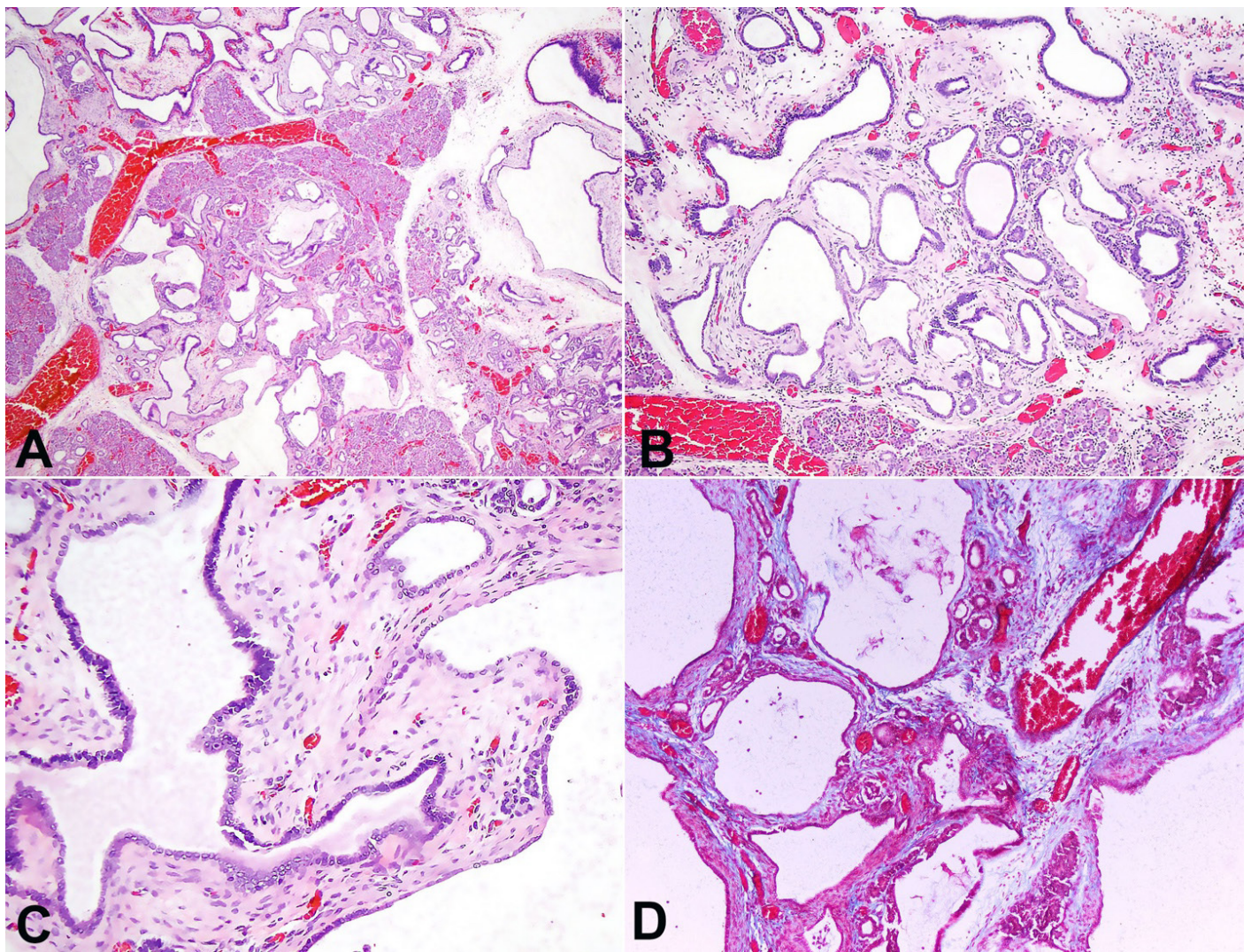


Figure 1. Photomicrography of the pancreatic hamartoma, showing in **A**, **B** and **C** – cysts and ducts lined by cuboidal epithelium, primitive mesenchymal proliferation and exocrine gland formation (H&E, 10X, 20X and 40X respectively); in **D** – mesenchymal proliferation by Masson's trichrome stain (20X).

The uninvolved pancreatic parenchyma showed normal exocrine and endocrine components, congestion and extramedullary hematopoiesis.

DISCUSSION

Pancreatic hamartomas are characterized as lesions with acinar, islet and ductal cellular components present in varying proportions and in a disorganized pattern. The exocrine and endocrine pancreatic tissue is well differentiated, and the ductal elements are irregularly branching or cystically dilated. The endocrine component may be seen as single cells or small group of cells interspersed within the exocrine component. Fibrous tissue, fat and primitive mesenchyme is often found in between these elements.¹⁻⁶

Pancreatic hamartomas are extremely rare tumors in adults but are even more so in children. Only a total of thirty-four cases of pancreatic hamartomas have been reported in the literature including twenty-seven in adults, five in children and two in newborns. Two of the five reported in children had Trisomy 18 and one adult had SAPHO syndrome, a chronic inflammatory disorder.¹⁻²³

In 1960, Smith et al.²³ originally described a pancreatic lesion in a two-month old girl with Trisomy 18 at the time of autopsy. The lesion was located at the tail of the gland, and was characterized by acinar and islet pancreatic tissue within a dense fibrous network and dilated ducts. In 1964, Rohde²² and colleagues reported a second pancreatic lesion in a Trisomy 18 child. In his case, a two-and-a-half-month girl was found to have an unusual pancreatic lesion described as having prominent immature connective tissue with sparse or absent acini, and distorted, often dilated ducts. These areas were surrounded by normal pancreatic tissue. The histologic description of these lesions is very similar to those seen in our case, and, despite the fact that these lesions were not labeled as hamartomas, their microscopic description seem to indicate that they may be considered hamartomas as well. Our case then, may be the third pancreatic hamartoma reported in association with Trisomy 18 Syndrome. Ectopic pancreas has also been reported in association with Trisomy 18.²²⁻²⁴ It is, therefore, recommended that careful examination of the pancreas be performed in individuals with Trisomy

18 to further characterize these lesions as one of the possible abnormal findings associated with Trisomy 18 Syndrome.

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