Case reports

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Antenatal detection of grossly distended bladder owing to absence of the urethra in a fetus with trisomy 18

SUMMARY An ultrasonic examination revealed a grossly distended fetal abdomen. Amniocentesis at 19 weeks' gestation showed raised amniotic fluid alphafetoprotein, a second band of specific acetylcholinesterase, and a fetal karyotype 47,XY,+18. The pregnancy was terminated and the necropsy examination showed absence of the urethra, grossly distended bladder, hydroureters, and congenital heart anomalies.

The routine antenatal use of ultrasound will occasionally reveal unexpected fetal abnormalities. In this report we describe a fetus with a large cystic abdominal swelling resulting from ascites and dilation of the bladder associated with trisomy 18 discovered on such an examination.

Case report

This 34-year-old woman had two previous pregnancies which led to normal offspring. There was no history of congenital abnormalities, chromosomal aberrations, or hereditary diseases. In her third pregnancy, at the ninth week booking visit, a real time ultrasound examination showed a normal fetus and gestational sac. At 18 weeks' gestation, a further routine ultrasound examination revealed abnormality of the fetal abdomen with apparent absence of the anterior abdominal wall. The abdomen was replaced by a very large cystic structure. The fetal skull had a BPD of 3.57 cm corresponding to 15½ weeks. It was difficult to see the lower limbs. One week later, another ultrasound examination confirmed the grossly distended fetal abdomen and alteration of fetal heart rate between 75 and 140 beats per minute. Transabdominal amniocentesis was performed, and 20 ml of clear amniotic fluid were withdrawn. The amniotic fluid alphafetoprotein (AFP) was 390 µg/ml (well above the 99.5 centile for 19 weeks' gestation). No rapidly adherent cells were observed after culture of amniotic fluid cells. Electrophoresis revealed an additional fast moving second band of specific acetylcholinesterase (AChE) characteristic of an open tube defect. The fetal karyotype was 47,XY,+18.

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The pregnancy was terminated at 20½ weeks' gestation using extra amniotic prostaglandin. At necropsy, the abdomen was grossly distended with marked thinning of the abdominal skin (fig 1). The abdominal distension was the result of ascites and a greatly distended bladder owing to absence of the urethra. The ureters were dilated and there was a mild hydronephrosis (fig 2). There was no neural tube defect. The heart was abnormal with a double outlet right ventricle with an overriding aorta. There was a ventricular septal defect and a hypoplastic left atrium and left ventricle. The fetus also showed a dolichocephalic skull, marked micrognathia, low set

FIG 1 Fetus at 20½ weeks showing features of trisomy 18 and gross abdominal distension with marked thinning of the abdominal skin.

FIG 2 Fetus at 20½ weeks with distension of the fetal bladder with hydroureters.

abnormal ears, and flexion deformity of the left wrist and of the fingers.

Discussion

Recently, a case similar to ours has been described¹ in which an ultrasound examination at 17 weeks' gestation showed a fetus with a grossly distended urinary bladder owing to an atretic urethra. This pregnancy was terminated and later the fetal karyotype was found to be trisomy 18 (47,XY,+18). The amniotic fluid AFP was normal, whereas in our case the amniotic fluid AFP was markedly raised. Raised amniotic fluid AFP levels, however, are not exclusively found in open neural tube defects, but have also been reported with several other fetal abnormalities,² such as congenital nephrosis, Meckel's syndrome, Turner's syndrome (cystic hygroma), gastrointestinal atresias, trisomy 13, and omphalocele/exomphalos. Nevin *et al*³ described a case at 27 weeks' gestation with a large distended bladder secondary to urethral atresia, other congenital abnormalities, and a raised amniotic fluid AFP. The raised amniotic fluid AFP in omphalocele/exomphalos and in distended fetal bladder owing to absence of the urethra is presumably the result of transudation of fetal serum through the markedly thinned fetal abdominal wall. The present case showed a second band of specific AChE characteristic of an open neural tube defect. A raised AFP and specific isoenzyme for AChE has been reported in a fetus at 17 weeks with a large exomphalos (part of Edwards's syndrome).⁴ The two bands in gel electrophoresis were very similar to those in NTDs. The UK Collaborative Acetylcholinesterase Study⁵ reported a positive gel AChE result in 47 of 63 (75%) pregnancies with a raised amniotic AFP which resulted in exomphalos.

On ultrasonic examination at 19 weeks' gestation there was marked variation of fetal heart rate and at necropsy the fetus had severe congenital heart anomalies. The gross ascites may have resulted from fetal heart failure. Although renal malformations are frequent in trisomy 18,⁶ absence or atresia of the urethra is rare. Our case is the third to describe the antenatal recognition of absence of the urethra in association with trisomy 18.^{1 4}

NORMAN C NEVIN*, J NEVIN*,
J MAURICE DUNLOP†, AND
MOYRA GRAY‡

*Department of Medical Genetics,
The Queen's University of Belfast;

†The Ulster Hospital, Dundonald, Belfast;
and ‡the Department of Pathology,
The Queen's University of Belfast,
Belfast.

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Requests for reprints to Professor N C Nevin, Department of Medical Genetics, The Queen's University of Belfast, Institute of Clinical Science, Grosvenor Road, Belfast BT12 6BJ.

Graves' disease and Down's syndrome

SUMMARY A case is described of severe thyrotoxicosis occurring in a patient with Down's syndrome, in which behavioural disturbance was a marked but reversible phenomenon. In addition, we report that antibodies capable of stimulating the thyroid were detectable in the sputum of both the patient and his mother.

Autoimmune disturbances including diabetes, adrenal failure, chronic active hepatitis, and particularly hypothyroidism are common in Down's syndrome.¹ However, few previous cases of hyperthyroidism have been described in such patients,^{2 3} and although on clinical grounds the majority of these patients probably had Graves' disease, the presence of immunoglobulins capable of stimulating the thyroid (TSI) has not previously been reported.

Case report

A 35-year-old man with Down's syndrome presented in April 1982 with a one-month history of weight loss, sweating, diarrhoea, heat intolerance, and difficulty in climbing stairs. In addition he had become increasingly irritable, withdrawn, and difficult to manage and had developed visual hallucinations. There was no significant past medical history and no family history of autoimmune disease.

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