Bilateral dysgerminoma and gonadoblastoma in association with XY gonadal dysgenesis

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Introduction

Pure 46 XY Gonadal Dysgenesis is a form of abnormal gonadal development. The high incidence of associated gonadoblastoma and subsequent dysgerminoma underline the importance of opportune diagnosis and management.

Case Report

An 18 year old woman presented with a history of primary amenorrhoea and hirsuitism. She had no galactorrhoea, and was otherwise well.

On examination, she weighed 62 kg, was 1.67 m high, and was normotensive (100/60 mmHg). There was an increase in hair growth on the upper lip, and the breasts were small. Abdominal examination was unremarkable. Pelvic examination confirmed scanty pubic hair, but otherwise normal external genitalia. The cervix looked normal, but the uterus was small. There were no adnexal masses.

Investigation results were as follows: serum total testosterone 5.4 nmol/l; LH 30.3 U/l; oestradiol level <50 pmol/l; prolactin 215 mU/l; TSH 0.8 mU/l; androstenedione 4.0 nmol/l; chromosome analysis 46 XY. An ultrasound examination of the pelvis revealed a small uterus. The ovaries could not be visualised. It was assumed that she had streak gonads and following counselling, the patient underwent laparotomy.

At operation, the uterus and tubes were small. There were gonads, in the normal ovarian position, each 2 cm in diameter and gritty. The pelvic peritoneum, omentum, kidneys and liver were normal. Bilateral gonadectomy was performed. Histological examination confirmed bilateral gonadoblastoma and dysgerminoma, invading the gonadal capsule on each side. No normal ovarian or testicular tissue was present.

Post-operatively, a CT scan of the abdomen and normal serum levels of the tumour markers âFP (<2KU/l) and ß-HCG (<2 IU/l), confirmed no evidence of metastatic spread. It was therefore decided that any potential benefit of adjuvant chemotherapy given to a teenager was outweighed by the possible risks, and no further treatment was given. Follow-up continues with further CT scanning and serum marker levels. The oral contraceptive pill has been prescribed to stimulate breast and uterine development. The hirsutism is improved.

Discussion

Forty six XY gonadal dysgenesis is a disorder of sexual differentiation. It is usually sporadic, but an X-linked recessive pattern of inheritance is recognised. The presence of a uterus, vagina and female external genitalia in this patient dates the dysgenetic process as prior to the sixth week of intra-uterine life. However, primary gonadal failure in such phenotypically normal females rarely presents prior to puberty; menarche may be delayed, or residual function in a dysgenetic testis may cause virilising changes e.g. hirsuitism. If karyotyping shows an XY pattern, testicular feminisation must be considered. In such patients, the uterus is absent and the vagina, blind-ending.

Patients with gonadal dysgenesis and a Y-chromosome have a high risk of gonadal malignancy, estimated to be 30% (Schellhas, 1974). Therefore prompt prophylactic gonadectomy is important. Tumour may be present despite normal clinical and ultrasound findings. In the presence of a uterus, future fertility is possible with hormone replacement and oocyte donation. Bilateral gonadectomy is therefore the preferred procedure, with adjunctive chemotherapy in advanced cases (Williams et al., 1990). Tumour markers e.g. âFP, HCG and LDH, may be used to monitor progress.

Due to the possible X-linked inheritance, all family members must be screened - the risk of gonadal malignancy extends to both phenotypically male and female siblings. Suitable counselling is of the utmost importance; in no way should a patient’s accepted sexuality be threatened.

References


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Rupture of an heterotopic pregnancy in the 15th week and normal continuation of the intrauterine gestation. Case report

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Summary

A rare case report of heterotopic pregnancy, ruptured in the 15th week of gestation is presented. The intrauterine pregnancy continued after the laparotomy and removal of the extrauterine fetus and ended with a normal delivery in the 39th week.

Key words: Heterotopic pregnancy; IVF.

Introduction

Combined intrauterine and extrauterine pregnancy is not a common occurrence in spontaneous pregnancy. However, the presence of simultaneous intrauterine and ectopic pregnancy among women treated by in-vitro fertilization (IVF) and embryo transfer is not uncommon [1, 2, 3]. The rupture of the extrauterine in the 15th week of gestation is a rare condition. The intrauterine (IVF) pregnancy that remained continued after the laparotomy and ended with a normal delivery.

Case Report

A 30 year old woman with an 8 year history of primary infertility due to severe endometriosis, conceived after the first IVF attempt. Ovarian hyperstimulation was induced using GnRH analogues (1.2 mg daily Suprefact Nasal spray, Buserelin, by Hoechst Germany), HMG (Metrodin, Pergonal by Serono, Aubonne Switzerland) and HCG (Pregnyl 10.000 IU i.m., by Organon, The Netherlands).

Twelve oocytes were aspirated and 5 embryos of good quality were transferred. The luteal phase was supported for two weeks with i.m. Progesterone (100 mg, Lutorm, Chropi). β-HCG 15 days post transfer was 349.6 IU/ml which on the 18th day rose to 16,970 IU/ml. Ultrasound in the 7th week revealed a single intrauterine viable embryo. Since both ovaries were hyperstimulated and enlarged no extrauterine pregnancy was suspected due to the existence of ascites in the Douglas space.

In the 15th week of gestation the patient presented at the Hospital with signs of intraperitoneal bleeding with acute abdomen, peritonitis and epigastric abdominal pain. Ultrasound revealed an ectopic embryo in the abdominal cavity as well as the existing intrauterine one.

After laparotomy the fetus and the placenta from the ruptured tube was found in the abdomen (Figure 1). After the operation ultrasound reassured the well-being of the intrauterine pregnancy. The histological report showed a fallopian tube with ectopic pregnancy. Fetus CRL was 75 mm.

The patient delivered a 3200 gr female normally in the 39th week of pregnancy.

Fig. 1. — Embryo in the 15th week of gestation with the ruptured fallopian tube and placenta.

Discussion

Heterotopic pregnancies seem to be presented more often since the introduction of Assisted Reproductive Technologies (ART).
In 1990 Molley et al. reviewed their assisted reproduction program and found 10 heterotopic pregnancies out of 1001 clinical pregnancies [1]. Heterotopic pregnancies should be suspected in most cases with high levels of β-hCG after ART, corresponding to more than one gestation. Especially in cases where more than 3 embryos are transferred, the ultrasonographer should suspect an ectopic pregnancy. The method of embryo transfer, the depth at which the catheter is inserted into the uterine cavity and the position of the patient after ET have also been mentioned by authors as possible factors for ectopic pregnancies [4, 5, 6].

In our IVF unit the incidence of ectopic pregnancies is 7.1% of all gestations while the incidence of heterotopic is 1.7%. Out of 1915 embryo transfers 461 pregnancies were achieved. Thirty three were ectopic and 8 heterotopic.

The case report presented here is a rare one because the rupture of the extrauterine pregnancy occurred in the 15th week of gestation. A careful follow-up of all IVF pregnancies, especially after transfer of high number of embryos is needed.

References


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