NEOPLASTIC DEGENERATION OF THE DYSGENETIC GONAD

G. GHIRARDINI, G. B. LA SALA, F. VALLI

Division of Obstetrics and Gynecology
Santa Maria Nuova Hospital
Reggio Emilia (Italy)

SUMMARY

A case of streak gonad syndrome 46, XY with dysgerminoma is reported. The case leads to discuss the most recent knowledges on neoplastic degeneration of the dysgenetic gonad. Pathogenetical significance of H-Y antigen in streak gonad tumors is stressed. A plan of diagnostic-therapeutic management in such cases is proposed.

Presence of streak gonads is the most important feature in numerous pictures of gonadal dysgenesis. In such cases it appears as justified the use of the term "streak gonad syndrome", instead of gonadal dysgenesis, as recently stressed by Bösze and Lazlo ([1]). These syndromes are usually diagnosed in peripubertal age during evaluations for primary amenorrhea, secondary amenorrhea and/or sterility. They can be subdivided into two groups: syndromes with presence of the Y chromosome or rearrangements of it and syndromes without the Y chromosome ([1, 2, 3, 4]). The second group encompasses pathologies where the karyotype shows partial or total monosomy or symmetrical rearrangements of the X chromosome ([2]) as well as defects of the X involving interbands q13-q24-27 ([5]). The same group also encompasses the "streak gonad syndrome 46,XX" and the "unilateral streak gonad syndrome" ([6]).

It has been established that only the streak gonad syndromes with Y chromosome or rearrangements of it are at high risk of neoplastic degeneration ([3]). This risk is increasing with age and about one third of these patients will have a gonadal tumor ([7, 8]). The more frequent tumors of the streak gonad syndromes are gonadoblastoma followed by dysgerminoma. Also cases of chorioncarcinoma, teratoma, teratocarcinoma, embryonal chorioncarcinoma and sinus endodermal tumor have been described ([7, 9]).

The gonadoblastoma is found to be bilateral in 36.5% of the cases, it is primitively benign ([9]). Not rarely develops from it a dysgerminoma, a malignant tumor of the germinal cells, which can substitute completely the gonadoblastoma. The dysgerminoma arises therefore from the gonadoblastoma, as malignant variant of it ([7]). It is sometimes difficult to differentiate between the two tumors, which are frequently mixed together.

Recent advances on H-Y antigen studies in sexual determination lead to be-

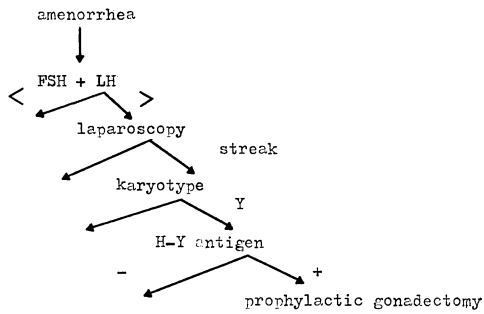
lieve that only in streak gonad syndromes with Y chromosome or rearrangements of it and H-Y antigen positivity is possible and unavoidable the neoplastic degeneration of the streak gonad ([8, 10, 11]). All but one ([12]) the reported cases of neoplastic degeneration of the streak gonad, when H-Y antigen has been determined, are H-Y positive, whereas no tumors have been described in H-Y negative streak gonad syndromes ([10, 11, 13]).

Müller and Siebers believe that in H-Y positive cases the streak would be consequence of a partial receptorial defect for the H-Y antigen. This would permit the presence of rudimentary testicular tissue, which would be the focus of neoplastic degeneration.

CASE HISTORY

A woman, 29 years old, married since 5 years was referred to our observation because of a pelvic mass. Past as well as family history of the patient were negative. Mammary and hair development at the age of 12. Primary amenorrhea (only withdrawal bleedings assuming estroprogestin medications). She was evaluated elsewhere for sterility and amenorrhea in 1978. A hypergonadotropinemia was found. There were no further evaluations. Patient's height 160 cm, weight 52 kg. Scanty axillary and pubic hair, mammary glands Tanner II. The gynecologic examination revealed hypoplastic vagina and uterus and presence of right adnexial mass (fetus' head at term) tender in consistence. Routine blood and urine examinations, as well as RIA determinations of PRL, HGH, TSH, T3, T4, T7, cortisol and testosterone were normal. LH and FSH levels were elevated (LH: 16 ng/ml, N.V. 0.5-5; FSH 30 ng/ml, N.V. 5). Also E2 determination revealed low values (20 pg/ml). The karyotype on peripheral blood with banding techniques (QFQ, RBA) was 46, XY. H-Y determination is now being made. At laparotomy a hyoplastic uterus with normal tubes was shown. The left gonad was transformed in a streak, whereas the right in a mass of 11 cm in diameter and of white-yellowish appearance, tender in consistence. Up today the patient is disease free. The histological examination of the left streak showed fibrous tissue with nodular hyperplasia of hilar cells and no germinal cells. The right gonad showed neoplastic tissue (dysgerminoma) with a great inflammatory reaction.

Table 1. — *Diagnostic-therapeutic plan in streak gonad syndromes.*



CONCLUSIONS

Recent advances in understanding pathologic conditions of sexual determination lead to a modern diagnostic and therapeutic management (table 1). Primary amenorrhea calls first for gonadotropin determination. In case of elevated levels of FSH and LH a laparoscopy should be done. In case streak gonads are present karyotype examination is unavoidable. In presence of Y chromosome or rearrangements of it H-Y determination is needed. If H-Y antigen is positive prophylactic gonadectomy should be done, to avoid neoplastic degeneration of the streak.

When H-Y determination will be more simple and further investigations will have confirmed the present results prophylactic gonadectomy will be no more necessary H-Y negative patients.

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