Seminoma in Marfan's syndrome

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Summary

A patient with a testicular seminoma and Marfan's syndrome is described. The association is unlikely to be by chance alone, and an explanation in terms of either an associated congenital defect, or a minor chromosomal anomaly, is discussed.

Introduction

The annual incidence of seminoma is 3.1 per 100 000. It is the most common tumour in males between the age of 20 and 34 (Twito and Kennedy, 1975). It is less common in Negroes and extremely rare in Bantu (Kaplan, Cohen and Roswit, 1951).

Marfan's syndrome, an inherited disorder of the mesenchyme, is clinically manifested by abnormalities of the eye, skeletal and cardiovascular systems. It is inherited as a simple Mendelian dominant; approximately 15% of cases are due to new mutations. The incidence of Marfan's syndrome is about 2/100 000 of the population.

A case is reported of a patient with Marfan's syndrome who was found to have a seminoma of the testis.

Case report

A 29-year-old white male, was found to have had abnormal stature in childhood. A definite diagnosis was not made and he was treated for flat feet. He was well until June 1977 when he started to lose weight. In November 1977, he became restless and was unable to sleep because of backache. By this time he had lost 6 kg in weight. The pain persisted, and in January 1978 he was found to have a grossly enlarged right testis and an abdominal mass. At the end of January 1978 he had a right orchidectomy and was referred to the Regional Oncology Unit for further investigation.

In his family history his mother, grandfather and sister have hyperextensibility of joints.

On examination, he was of anxious disposition, normal intelligence, but his appearance was remarkable (Fig. 1). He had arachnodactyly (Fig. 2) outward displacement of the chest (pigeon breast), dolichocephaly and a high arched palate. Although he had kyphosis there was no scoliosis. Hyperextensibility

FIG. 1. The patient

of the joints was particularly marked in the knees. He had a divergent squint and severe myopia, but there was no ectopia lentis. His height was 180 cm and the ratio of the upper segment (pubis to vertex) to the lower (pubis to sole) was 0.84 (normal ratio

in Whites being 0.92 ± 0.04). His arm span was 194 cm. A systolic ejection click was audible but no other abnormality of the cardiovascular system was found. A hard mass $10 \text{ cm} \times 5 \text{ cm}$ was noted in the hypogastrium. A small rather hard remnant of the right spermatic cord was present in the scrotum whereas the left testis felt normal.

FIG. 2. The patient's hands showing arachnodactyly.

On investigation his chest X-ray was normal. An IVP showed a functionless right kidney but a normally functioning left kidney which had undergone compensatory hypertrophy. Serum urea was 7.5 mmol/l , serum creatinine $132 \text{ } \mu\text{mol/l}$ and creatinine clearance 32 ml/min . Histology of the testicular tumour showed a seminoma of conventional type, although in some areas the features approached those of a spermatocytic seminoma. The tumour involved the spermatic cord and there was evidence of venous and lymphatic infiltration. Serum α -feto-protein was normal. Serum human chorionic gonadotrophin level was greater than 50 ng/l .

Chromosomal studies using a banding technique showed a normal genotype.

Discussion

To the authors' knowledge this is the first case of seminoma in association with Marfan's syndrome. This may be just a coincidence of a rare tumour of germinal origin occurring with a rare syndrome of mesenchymal origin. However, the prevalence of Marfan's syndrome and of a seminoma of testis makes the chances of this occurring $6/10^{10}$, a remote possibility.

Genetic factors predisposing to neoplasm may be involved. Many congenital malformations are associated with neoplasms, particularly in childhood (Bolande, 1977). Testicular tumours occur 15 to 40 times more commonly in undescended testes than in intrascrotal testes. About $1/80$ inguinal testes, and $1/20$ abdominal testes may be expected to develop neoplasia (Campbell, 1942). The most common tumour that develops is seminoma. It is suggested that congenital imperfection of the testes, rather than its abnormal site is the predisposing factor (Sohval, 1956).

The concept of cancer running in families is well documented (Anderson, 1978). Of 2336 proved or provable single gene traits, 8.6% have neoplastic tendencies (Mulvihill, 1977). These include the so-called 'chromosome instability syndromes' such as Fanconi's anaemia, xeroderma pigmentosa, the naevoid basal cell carcinoma syndrome, and incontinentia pigmenti (Hecht and McCaw, 1977). Thus chromosomal anomalies in the host, as opposed to abnormal chromosome patterns found in certain tumours (Jackson, 1978) may be associated with an increase in cancer risk. In particular, abnormal sex chromosome patterns, usually in phenotypic females, are associated with dysgerminomas, histologically identical to the seminoma of the male (Scully, 1977). Seminoma itself has been reported in association with Klinefelter's syndrome (Isurugi *et al.*, 1977).

Associated congenital anomalies, or minor chromosomal aberrations, undiagnosed by present techniques, may be responsible for carcinogenesis in this case.

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