Catégorie

"Case Report Abstract"

Genetic predisposition to breast cancer occurring in a male-to-female transsexual patient

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Introduction

Breast cancer affects less than 1% of all male cancer patients. In 5-20% of cases, genetic predisposition is involved, mostly due to mutations of the BRCA2 gene. Hormonal imbalance between oestrogens and androgens is another predisposing factor. Male-to-female (MtF) transsexual patients usually undergo long-term cross-sex hormone therapy, which could expose them to higher risks of developing hormonally-dependent cancers.

We report the case of a MtF transsexual patient diagnosed with breast cancer. A pathogenic BRCA2 mutation was found in the patient and her family.

Case report

Diagnosed with sexual identity dysphoria, the patient sought endocrine treatment at the age of 46 in order to start hormonal therapy. For 7 years, the patient was administered anti-androgens associated with oestrogens. The physical transformation was found satisfactory by the patient and the treatment was followed unremarkably. After 7 years, a routine mammography revealed a suspicious region with microcalcifications on the right breast. Biopsy was performed and the analysis found a high grade ductal in situ carcinoma without obvious signs of infiltration. Hormone therapy was interrupted and the patient underwent right mastectomy. A focally undifferentiated ductal carcinoma was found, oestrogen and progesterone-receptors positive. 2 years later, local recurrence was diagnosed on the mastectomy scar.

Genetic analysis revealed the heterozygous c.9117G>A mutation of the BRCA2 gene. The same mutation was known in a kindred bearing the same family name as our patient and living in the same region.

Conclusion

This is the first report of breast cancer occurring in a MtF transsexual patient with a proven genetic abnormality. It raises awareness of the particular care required by MtF patients due to their modified hormonal environment.

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