An Unexpected Diagnosis of Turner Mosaicism: A Pregnancy-related Clinical Challenge

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INTRODUCTION

Turner Syndrome affects 1/2000 female births with cardiac anomalies seen in 5-50% of these cases. Women with Turner Syndrome are known to have primary ovarian insufficiency and are at increased risk of congenital heart defects, ischemic heart disease, hypertension, stroke and aortic dissection.

METHODS

An evaluation of a pregnancy complicated by maternal Turner mosaicism with severe maternal vascular malformation and secondary hypertension leading to a preterm delivery.

CASE PRESENTATION

Here we present the challenging case of a 35-year-old primagravida with a medical history of congenital coarctation of the aorta with distal hypoplasia and associated with an extensive collateral system supplying the lower half of the body with significant renal impairment. Although strongly advised against pregnancy, spontaneous conception occurred. A dating scan showed increased nuchal translucency, and subsequent chorionic villus sampling confirmed no major trisomies or X-chromosomal aneuploidies. Increased nuchal translucency along with maternal anatomical malformations prompted consideration of a maternal genetic disorder, and subsequent maternal karyotyping surprisingly confirmed mosaicism for Turner syndrome.

An elective cesarean section was performed by a multidisciplinary team at 28 weeks gestation due to maternal complications, following extensive MRI and doppler mapping of the pelvic vessels.

DISCUSSION

Women with Turner syndrome have increased risk of cardiac malformation, hypertension, ischaemic heart disease and stroke1. It has been reported that up to 20-25% of women with Turner syndrome have a associated cardiac malformation2, and it is estimated that of all aortic dissections in women, 50% occur during pregnancy3.

As we know, stroke volume increases throughout pregnancy with increased resting pulse rate and increased overall blood volume in the third trimester. These changes, along with acute cardiovascular changes during labour, may cause further cardiovascular compromise in women with underlying cardio-vascular disease. Such factors must be considered, discussed and investigated during preconseptual counseling in women with Turner syndrome.

REFERENCES

