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TUS. Turner's Syndrome

NIHR BioResource – Rare Diseases study project

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Summary

Turner syndrome (TS) is a multisystem syndrome affecting women and girls, arising due to the partial or complete absence of one X chromosome. It occurs in approximately 1 in 2,500 liveborn girls. Features of the syndrome include failure to go through puberty, short stature, lymphoedema, renal malformations, hypothyroidism, intellectual impairment and cardiovascular abnormalities.

The cardiovascular complications of TS may be life-threatening. Around 7% of girls with Turner syndrome have coarctation of the aorta, and around 15% have bicuspid aortic valve. In later life aortic dissection may be a catastrophic complication. TS women are at much higher lifetime risk of aortic dissection compared to the general population. Decisions regarding when to operate on a dilated aorta in TS are complex and based on relatively sparse evidence. In addition, the later-life risks of hypertension, obesity and metabolic syndrome/diabetes are significantly increased.

The natural history of the development of aortic pathology is unknown, as are the molecular events giving rise to the aortopathy. The relationship between X-chromosome genotype and cardiovascular risk is incompletely understood, and the potential role of variants elsewhere in the genome is unknown. More evidence from clinical and genetic studies is required to improve the clinical care of TS women, particularly regarding cardiovascular risks.

Recruitment Criteria

Inclusion

Women and girls of any age can be recruited to the TUS project if they have been clinically diagnosed with Turner Syndrome and karyotyping has identified an aneuploidy consistent with the diagnosis.