An improved understanding of pregnancy in women with Turner's syndrome may save lives! BJOG-21-1221

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Around 25% of maternal deaths are related to heart disease and up-to 20% of these are the consequence of aortic dissection (Lameijer et al, Neth Heart J (2020) 28:27–36). Connective tissue disorders (e.g. Marfan's, Loeys-Dietz, Ehlers Danlos), Bicuspid aortic valve, Turner's Syndrome (TS) and pre-existing coarctations constitute the bulk of aortopathies encountered in pregnancy. Although a true XO karyotype in pregnancy had been rarely encountered historically due to the reduced fertility in these patients, women with a mosaic TS may have normal fertility and in recent years reproductive technologies with oocyte donation have increasingly been used in the sub-fertile TS population. Although pregnancies in such women remain infrequent, the risk of death during pregnancy amongst all women with TS (inclusive of mosaicism) has been reported as high as 2%, 150-200 times greater than the general population. This increased risk has led to the establishment of guidelines for the use of reproductive technologies in these women (Karnis. Fertility Sterility (2012) 98(4),787-91).

The rarity of these cases means that available data, even when drawn from multicenter registries, has been relatively sparse. A study of TS patients gathered from ten cardiovascular centres over 12 years evaluated 68 pregnancies (Grewal J, et al. Heart 2021;107:61–66). Although the majority had no structural cardiac disease and no major cardiovascular complications were observed, the adverse obstetric event rate was around 20% with a similar rate for adverse fetal outcomes. Like the present paper (BJOG-21-1221) which reports a larger population, these cases were followed within centres offering a dedicated obstetric-cardiac clinic.

Standardized guidelines for preconceptual and perinatal care of these women have improved outcomes, as evidenced by a French study evaluating cohorts from before and after the establishment of such guidance in France (Cadoret et al, EJOGRB(2018) 229,20–25). Whilst these guidelines encourage the care of TS mothers in dedicated obstetric-cardiac centers, this is not always the case; either due to lack of diagnosis of the condition (esp in mosaic cases with milder phenotypes) or lack of appreciation of the risks. The present paper reports only cases followed in cardiac-obstetric centres; without an overall population pregnancy case number it will remain uncertain what proportion of women with TS were followed in an appropriate centre and whether the reported cohort therefore excludes cases with worse outcomes.

Although it might be expected that mothers followed within appropriate centres receive optimal care, studies continue to show that this is not always so. In the present cohort only half of the women had a pre-pregnancy cardiac evaluation and 20% had no record of imaging during pregnancy; perhaps reflecting late referral to the tertiary centre. While this remains a high-risk population with risks for both maternal and fetal adverse outcomes with high haemorrhage rates, hypertensive disease rates (˜19%) and an SGA rate exceeding 20%, the data from this cohort is reassuring with respect to severe perinatal morbidity. Particularly striking in this cohort was a caesarean rate of 67%.

To provide women with TS optimal care, preconceptual evaluation and pregnancy planning with medical optimisation is vital, with subsequent close follow-up in an obstetric-cardiac centre capable of managing whatever complications this cohort might experience, even if rare.

Disclosure of Interest

RB has no conflicts of interest

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