

## **Turner karyotype and childbirth**

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Turner syndrome (TS) is a sex chromosome aberration and occurs in 1/2500 live born girls. One X chromosome is absent or structurally changed in all (monosomy) or some (mosaicism) of the cells. TS is characterized by short stature, ovarian failure and cardiac defects. Mortality is increased, mostly owing to cardiovascular disease and aortic dissection. Pregnancies in women with TS are rare, but have increased owing to oocyte donation (OD) and are reported to be of high risk.

*The aim* of this thesis was to describe characteristics of mothers to girls with TS and characteristics of newborns with TS and to evaluate the obstetric and neonatal outcomes in women with Turner karyotype and whether the pregnancy increased morbidity after delivery.

Characteristics of mothers giving birth to girls with TS from 1973-2006 and their newborns with TS were investigated in a study using the Swedish Genetic Turner Register (SGTR) and the Swedish Medical Register (MBR). Mothers to girls with TS were older and shorter than mothers from the general population. More girls with TS were born late preterm and were small for gestational age than controls. In a registry study, using the SGTR, the MBR, and the Cause of Death Register (CDR), 115 women with Turner karyotype who gave birth to 208 children (after both spontaneous and OD pregnancies) born 1973-2007, were studied. One woman had an aortic dissection. Singletons of women with Turner karyotype had lower gestational age, but similar size at birth and the rate of birth defects did not differ. In a Nordic, descriptive study on women with TS who had delivered after OD 106, women and 131 children born from 1992-2011 were included, and data from medical records were registered. The rate of hypertensive disorders during pregnancy was 35%. Life-threatening events occurred in four pregnancies (3.3%) including one with aortic dissection. The rate of preterm birth was 8% and low birth weight 9%. In a population-based registry study, mortality and morbidity in 124 women with Turner karyotype who had given birth from 1973-2010 was compared with women with Turner karyotype without childbirth and a control group from the MBR. The SGTR, the MBR, the National Patient Register, the CDR and Cancer Register were used. Morbidity and mortality in the total Turner group were increased as compared with the controls. Morbidity from cardiovascular diseases was increased before and during pregnancy but similar after more than one year after delivery and no deaths were seen.

*In conclusion*, pregnancies in women with TS are high risk pregnancies owing to hypertensive disorders and aortic dissection. Neonatal outcomes in women with TS are generally reassuring. Women who gave birth to girls with TS were shorter and older.

**Key words:** Turner syndrome, obstetric, neonatal outcome, Turner karyotype, pregnancy, maternal, neonatal characteristics.

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