The Prevalence And Outcome Of Sex Chromosome Abnormalities Detected Prenatally In Scotland

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Introduction:

Prenatal diagnosis (PND) via amniocentesis or chorionic villus sampling may result in the identification of a sex chromosome abnormality, often as an incidental finding.

Aims:

To ascertain the incidence of sex chromosome abnormalities detected by prenatal diagnosis in Scotland and to determine the characteristics and outcomes of these cases.

Methods:

A retrospective review of all prenatal karyotypes performed in Scotland between 2000 and 2012 was conducted. Data linkage was performed to obtain information regarding maternal characteristics, pregnancy outcomes and outcomes for affected infants. More detailed outcome data were available for 2 regions of Scotland: the Grampian and West of Scotland areas.

Results:

Over the 12 years, 28,145 karyotypes were performed in Scotland. Indications for PND are demonstrated in Figure 1. Records were available for 27,152 (96%) of these pregnancies. Karyotype abnormalities were identified in 8% of tests performed, with sex chromosome abnormalities being identified in 1% of all tests performed. Figure 2 demonstrates the results of all abnormal karyotypes. The commonest sex chromosome abnormality was 45,X.

A total of 126 cases of sex chromosome abnormalities were identified prenatally in the West of Scotland and Grampian areas. Of these, 54 (43%) progressed to live birth; 4 (4%) resulted in intrauterine death or stillbirth and 49 (46%) resulted in termination of pregnancy. 71% of pregnancies which progressed to live birth received genetic counselling vs 2% of those which progressed to termination of pregnancy. Of the 54 live births, 19 (35%) of the infants were clinically reviewed by endocrinology during the study time period. Only 11/54 (20%) in the West of Scotland/Grampian areas had undergone an endocrine review as demonstrated in Figure 3.

Conclusions:

Sex chromosome abnormalities are identified in approximately 1% of all pregnancies undergoing a prenatal diagnosis. There is a need for an improved structured pathway for prenatal as well as postnatal care of the mother and the offspring.

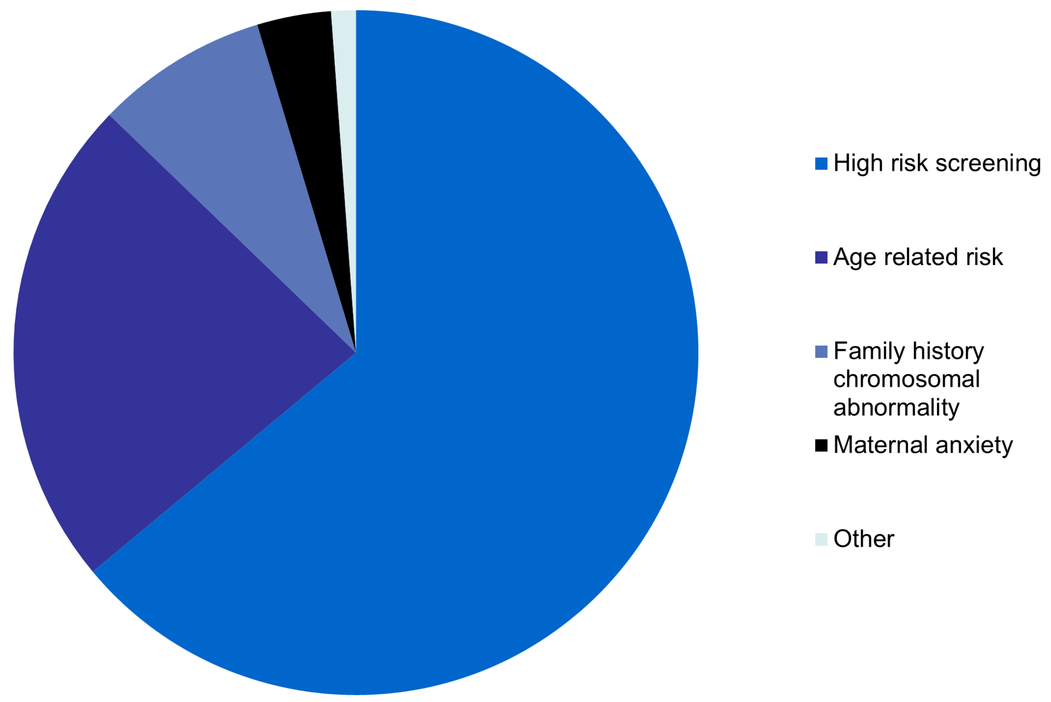

Figure 1. Indications for amniocentesis/CVS

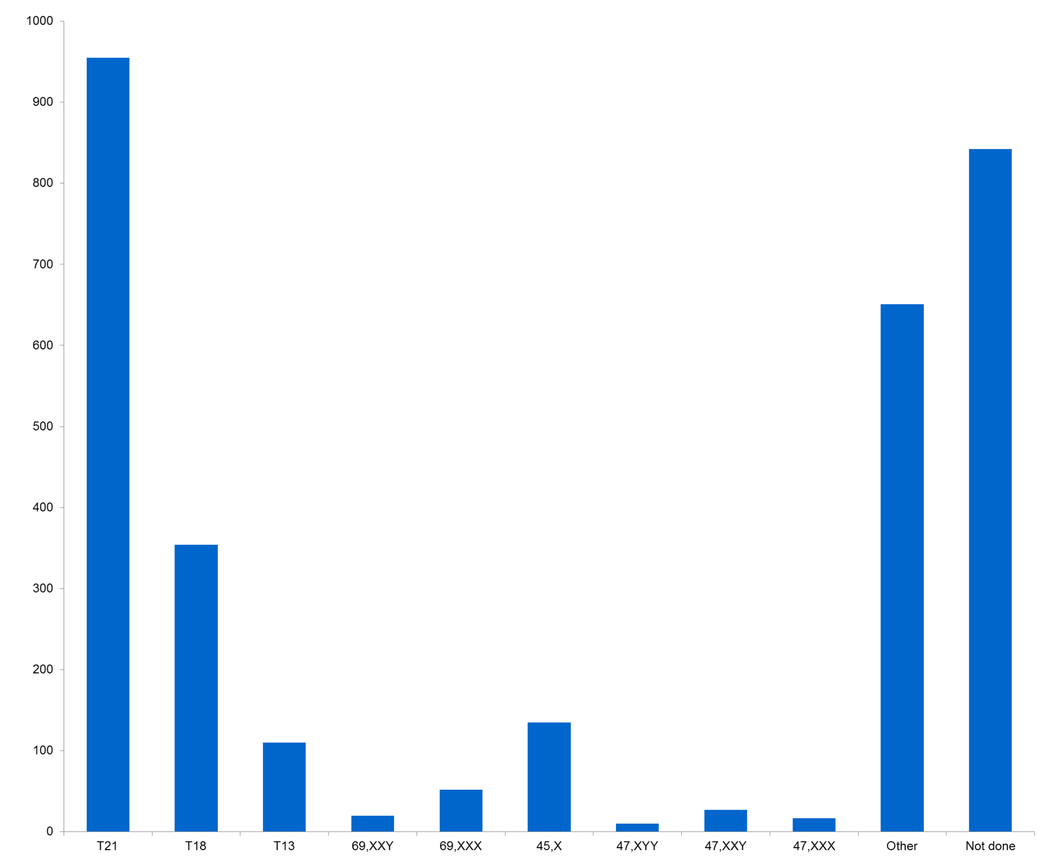

Figure 2. Outcomes of abnormal karyotypes

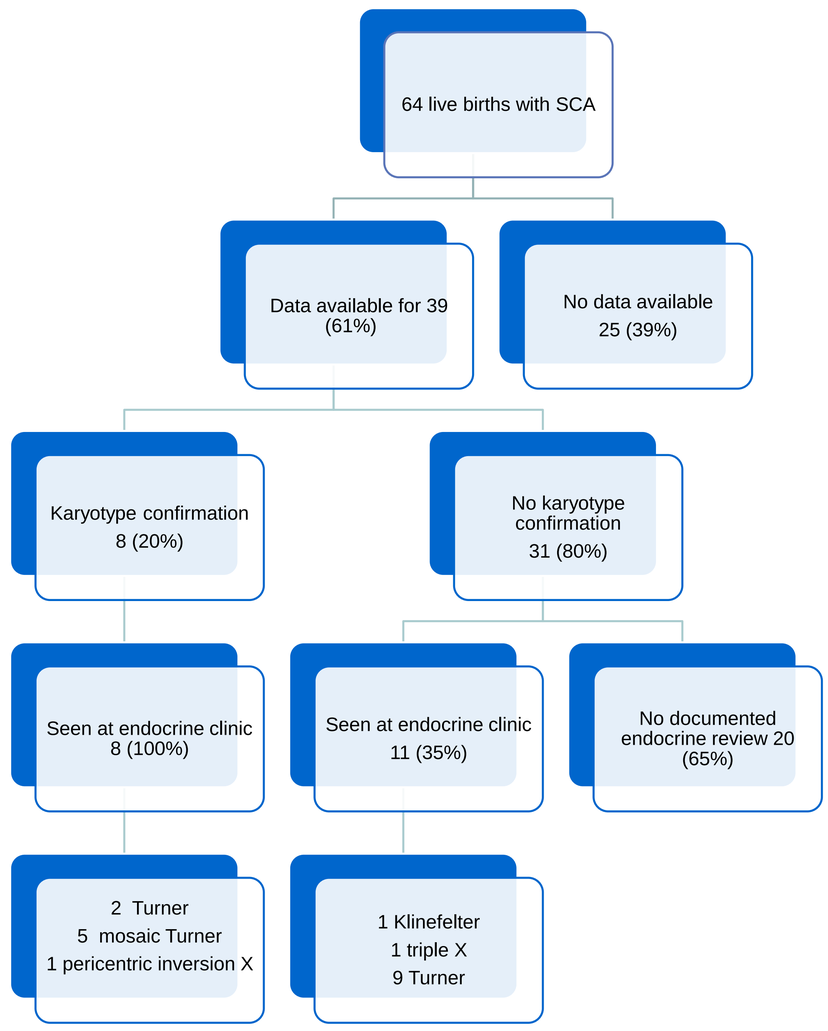

Figure 3. Outcomes of live births with prenatal diagnosis of SCA in Glasogw and Grampian