Prenatal diagnosis of sex chromosome abnormalities

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Objective

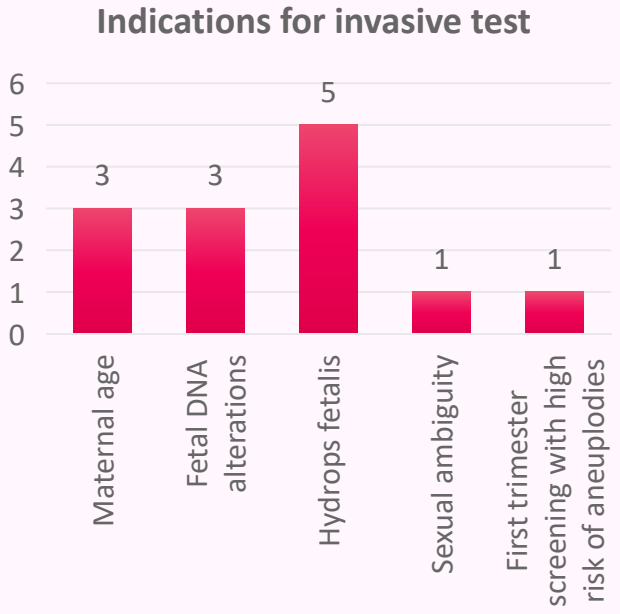
Sex chromosome abnormalities include 45, X0 (Turner syndrome), 47 XXX (Trisomy X), 47 XXY (Klinefelter syndrome) and 47 XYY (double Y syndrome). The aim of this study was to evaluate the cases with prenatal diagnosis of sex chromosome abnormalities and the outcomes of the pregnancy.

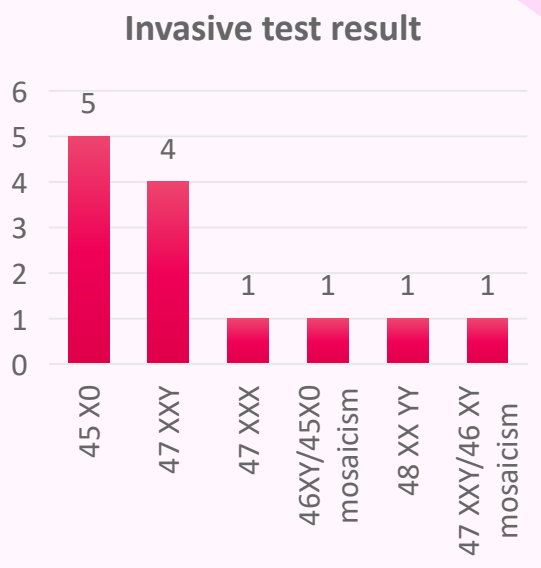
Methods

We conducted a retrospetive study where we analysed the data of cases with prenatal diagnosis of sexual chromosome abnormalities. Our analysis included the cases that ocurred in our hospital between 2014 and 2022. Demographic data, indications for invasive testing, ultrassound features and outcomes were obtained.

Results

Our evaluation included 13 cases.
Mean maternal age was 32 years old.
Mean gestagional age at diagnosis was 15 weeks (11 - 22 weeks).
2 chorionic villus sampling and 11 amniocentesis were performed.





Summary of the ultrasound findings and outcomes according to the invasive test result

Invasive testing result	Ultrasound features	Fetal or neonatal outcomes	Follow up or Foetopathology
45 X0 (*n=5*)	Hydrops fetalis (*n=5*) Cardiac malformation (*n=2*)	Fetal death (*n=1*) Legal termination of pregnancy (*n=4*)	Hydrops fetalis (n=5) Phenotypic changes related to 45 X0 karyotype (n=5) Renal dysplasia (n=2) Cardiac malformation (n=2) Aortic coarctation (n=1) Omphalocele (n=1)
47 XXY (*n=4*)	None (*n=3*) Unknown (*n=1*)	Live newborn (*n=3*)	Healthy
		Legal termination of pregnancy (*n=1*)	Hypogonadism, hypospadias
47 XXX (*n=1*)	None (*n=1*)	Live newborn (*n=1*),	Unknown
46 XY/45 X0 mosaicism (*n=1*)	Sexual ambiguity (*n=1*)	Live newborn (*n=1*)	Left cryptorchidism
48 XX YY (*n=1*)	Unknown (*n=1*)	Legal termination of pregnancy (*n=1*)	No morphologic changes
47 XXY/46 XY mosaicism (n=1)	None (*n=1*)	Live newborn (*n=1*)	Healthy

Conclusion: The most prevalent sex chromosome abnormalities were the 45 X0 (Turner syndrome) and 47 XXY (Klinefelter syndrome). It is shown the importance of prenatal diagnosis of sex chromosome abnormalities, not only to evaluate the possibility of termination of the pregnancy (in the most severe cases), but also for postnatal follow up.