

# Cytogenetic study of Sex chromosomal abnormalities in Egyptian DSD patients

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## OBJECTIVES

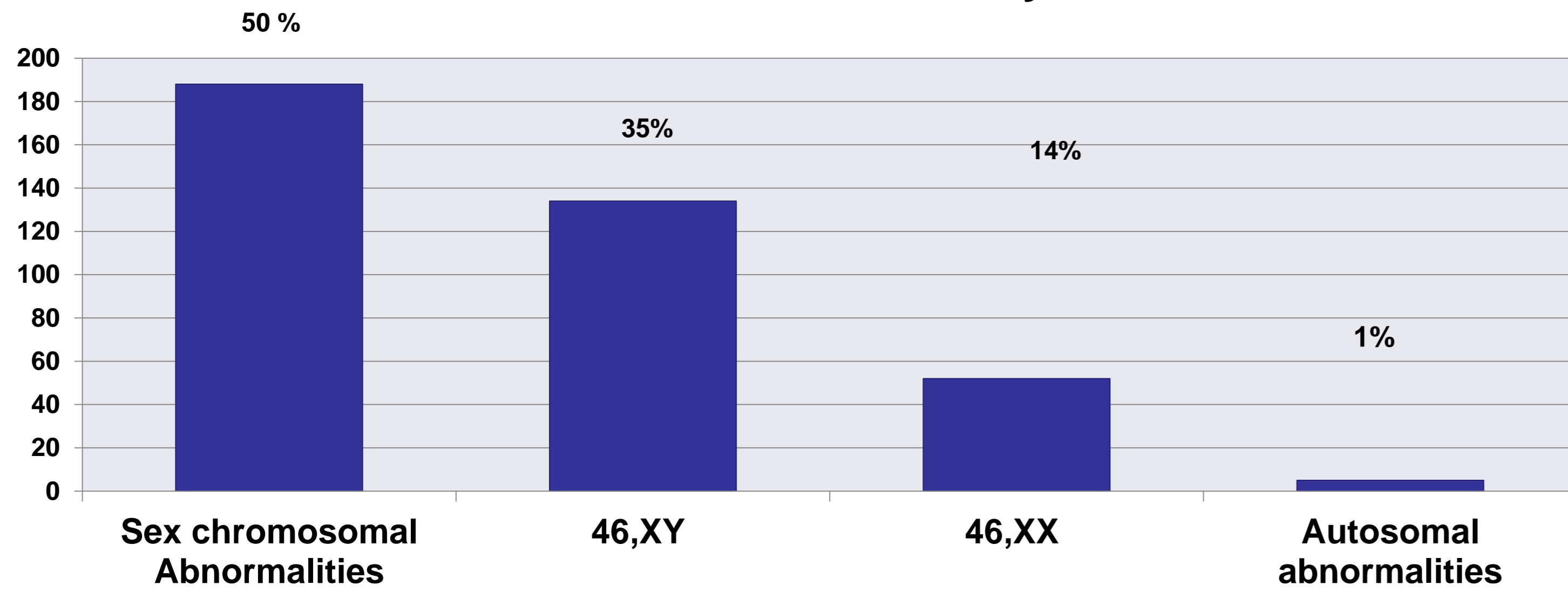
The study included 379 patients comprising a wide spectrum of presenting features, associated with different arrays of chromosomal abnormalities aiming at studying the prevalence of Sex chromosomal abnormalities among DSD patients

## METHODS

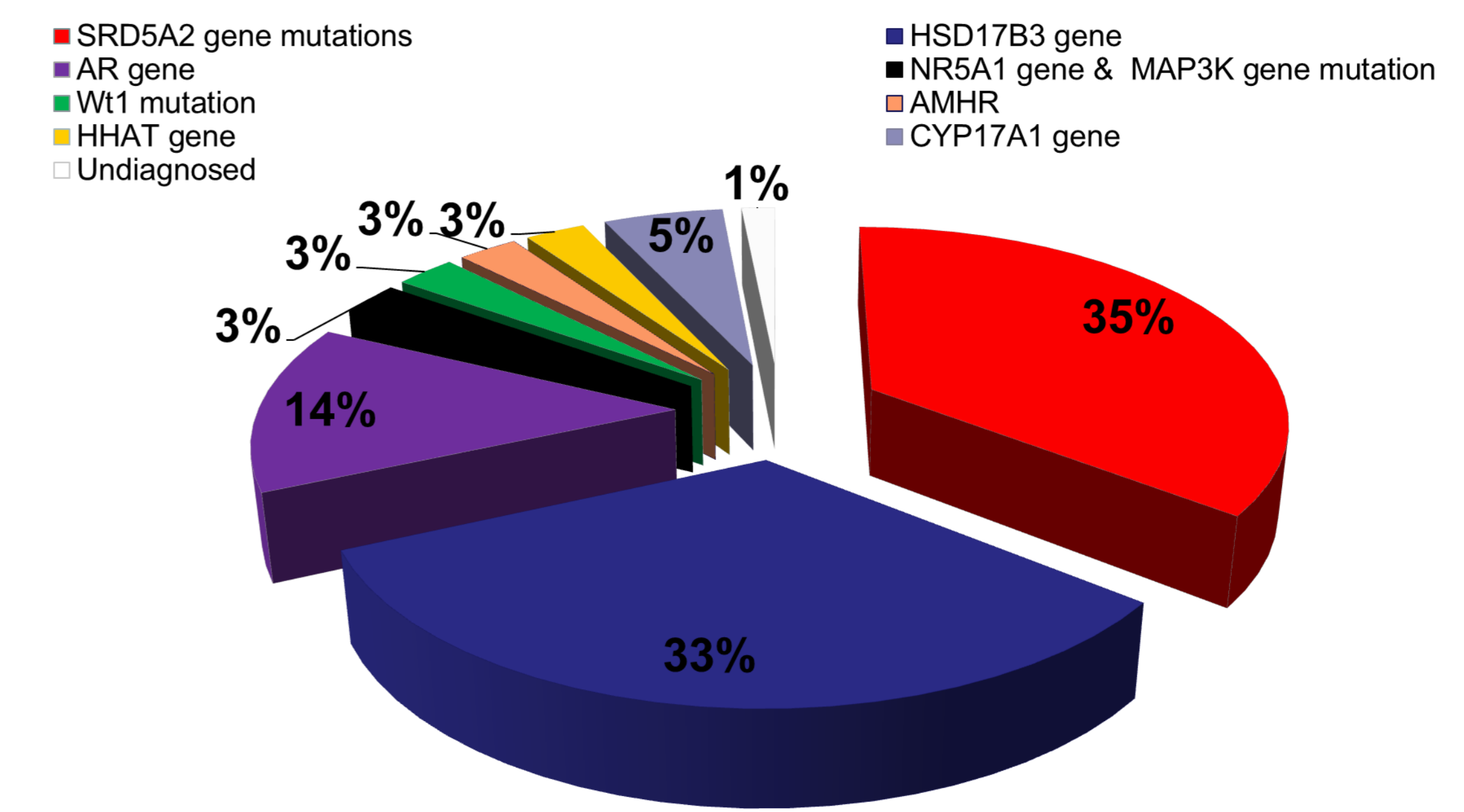
Patients were subjected to detailed clinical examination, pubertal staging, cytogenetic and FISH analysis. Laparoscopy with gonadal biopsy and FISH on gonadal tissue cells were done when indicated

## RESULTS and GRAPHS

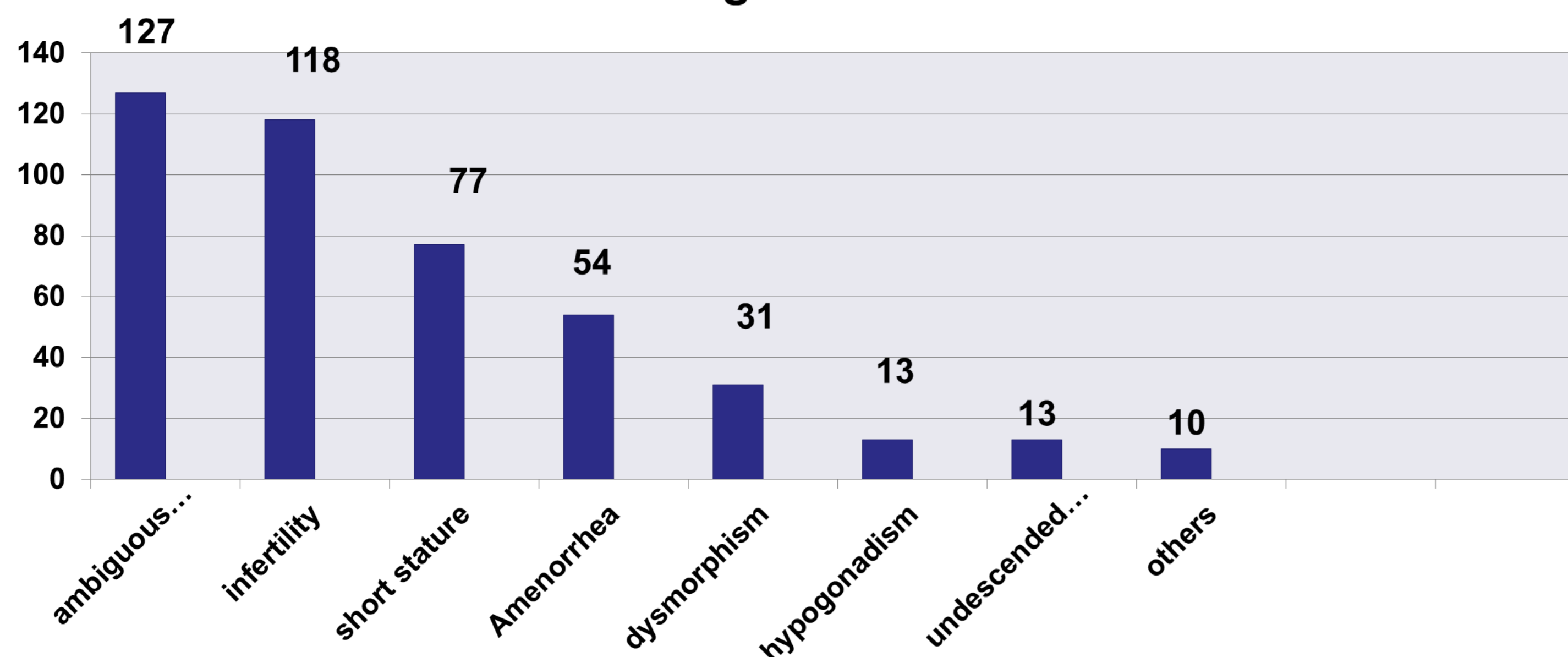
Results of Chromosomal Analysis



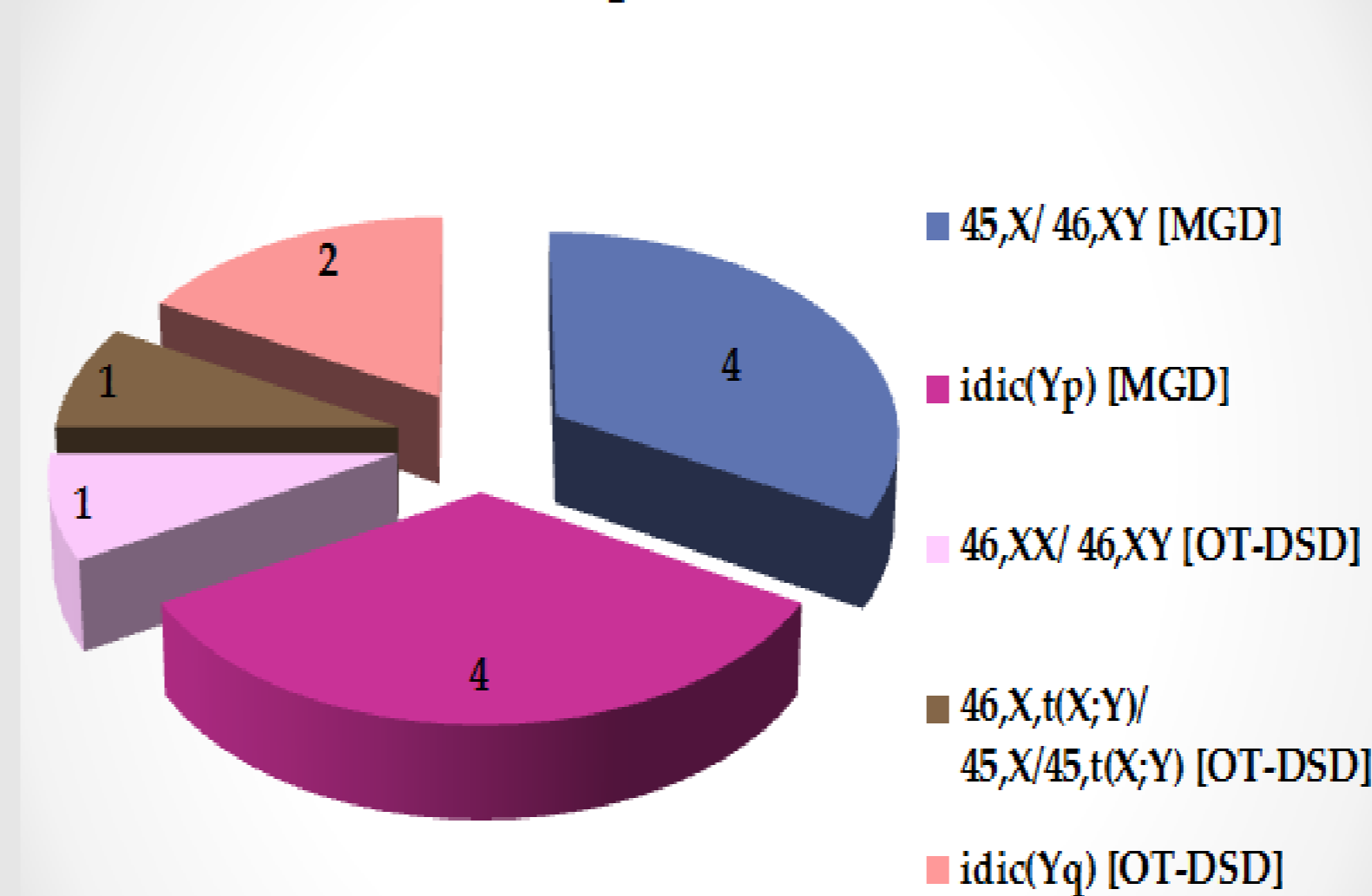
Monogenic causes detected among studied patients



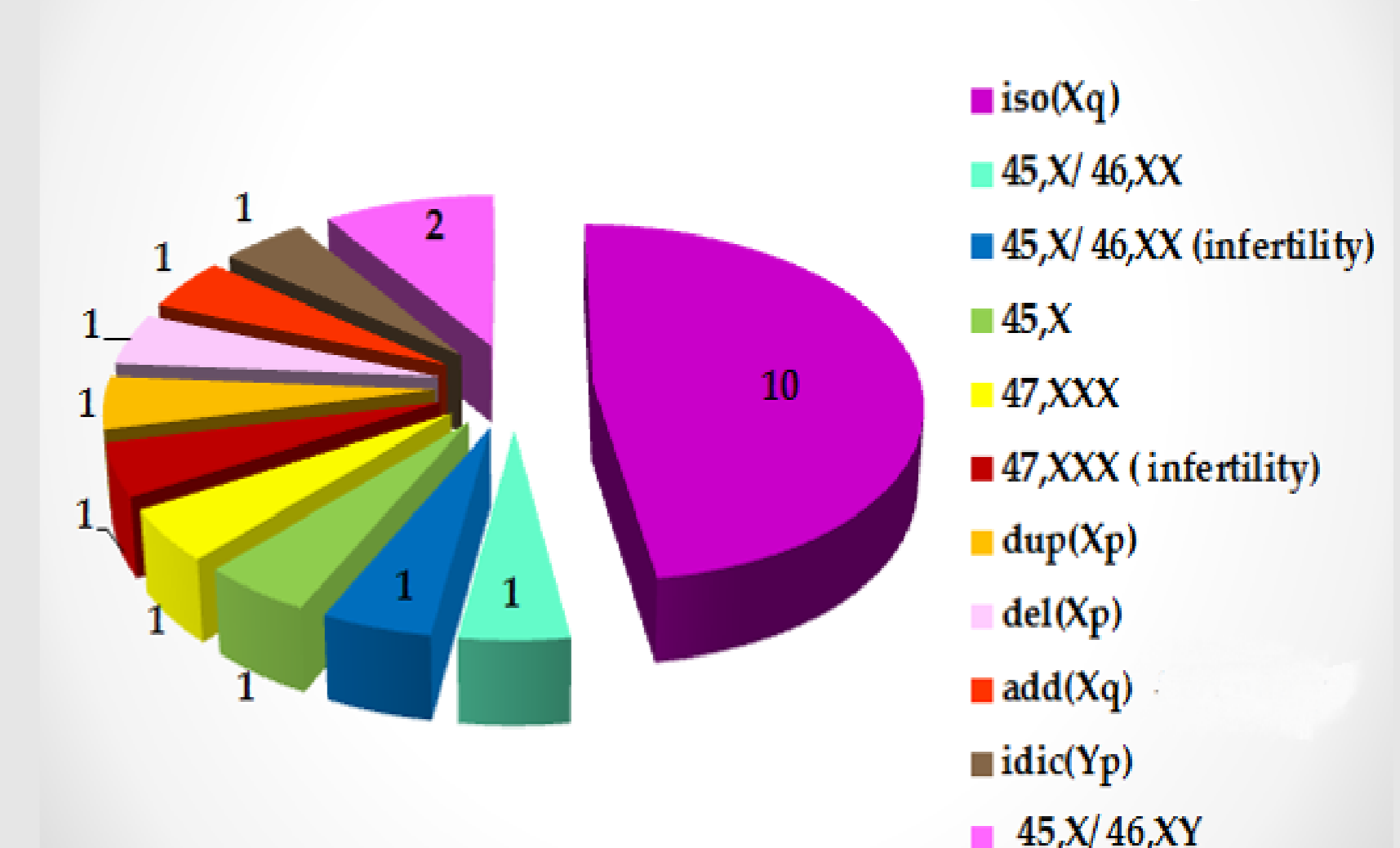
Presenting features



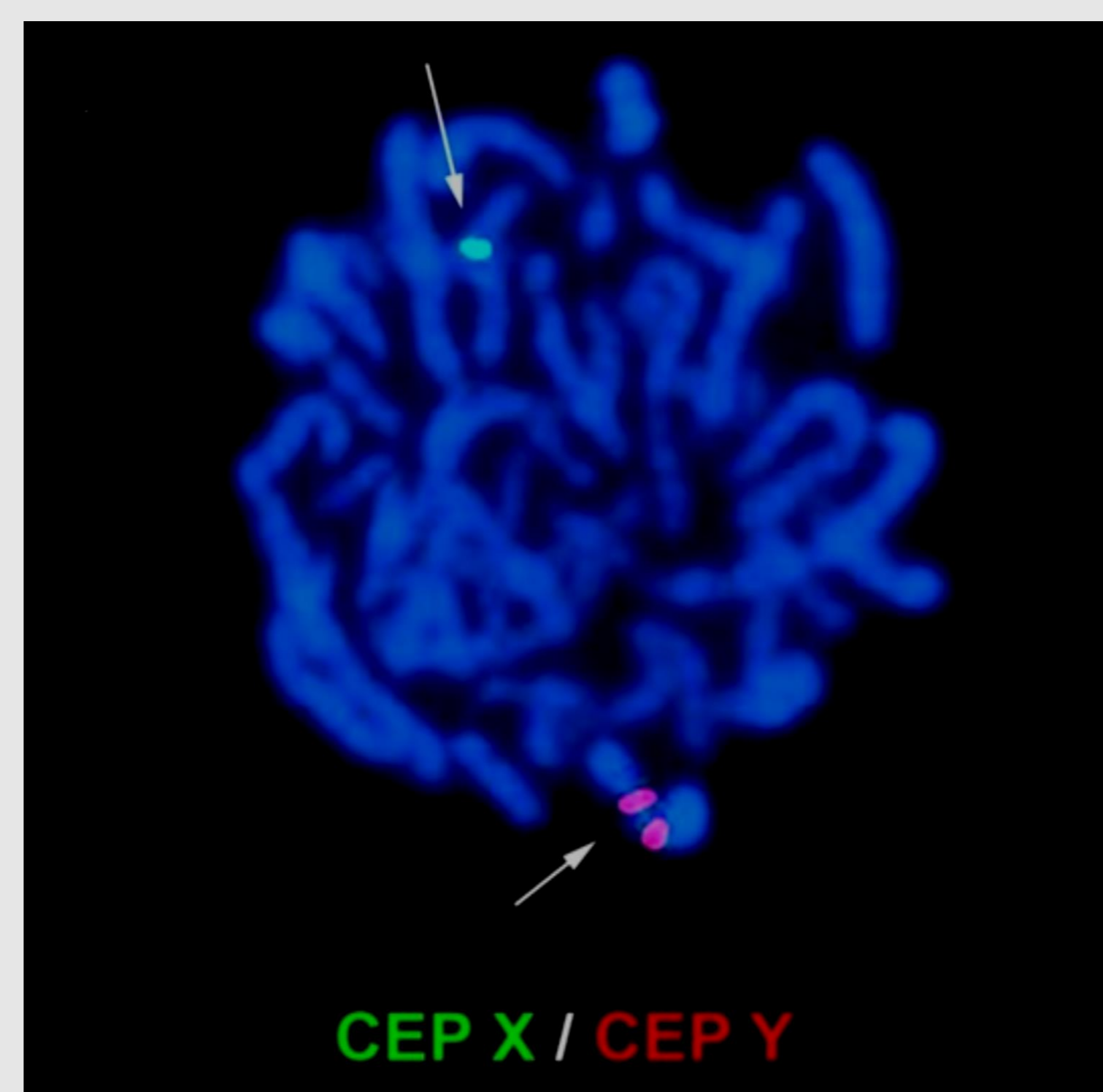
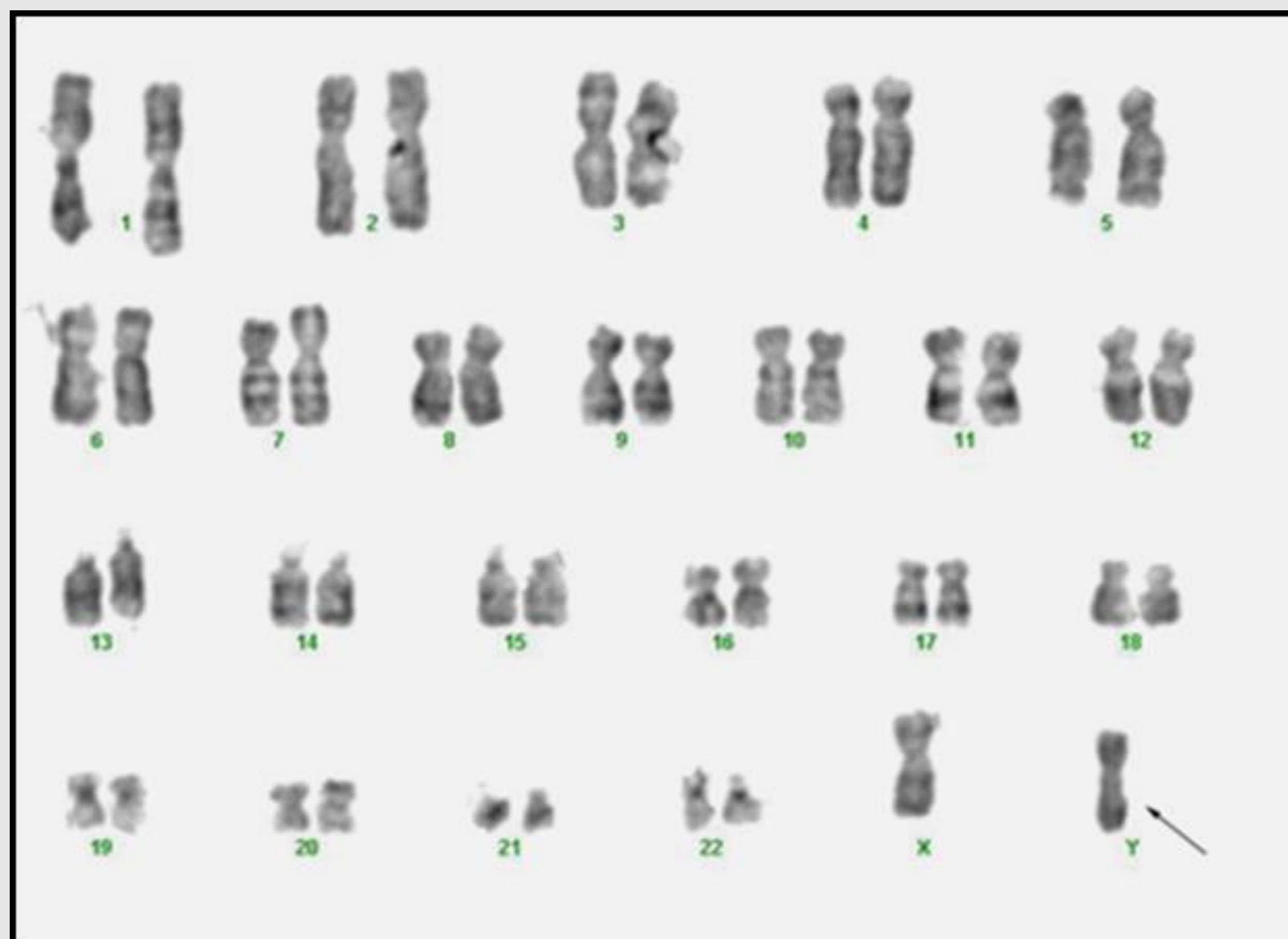
Ambiguous Genitalia



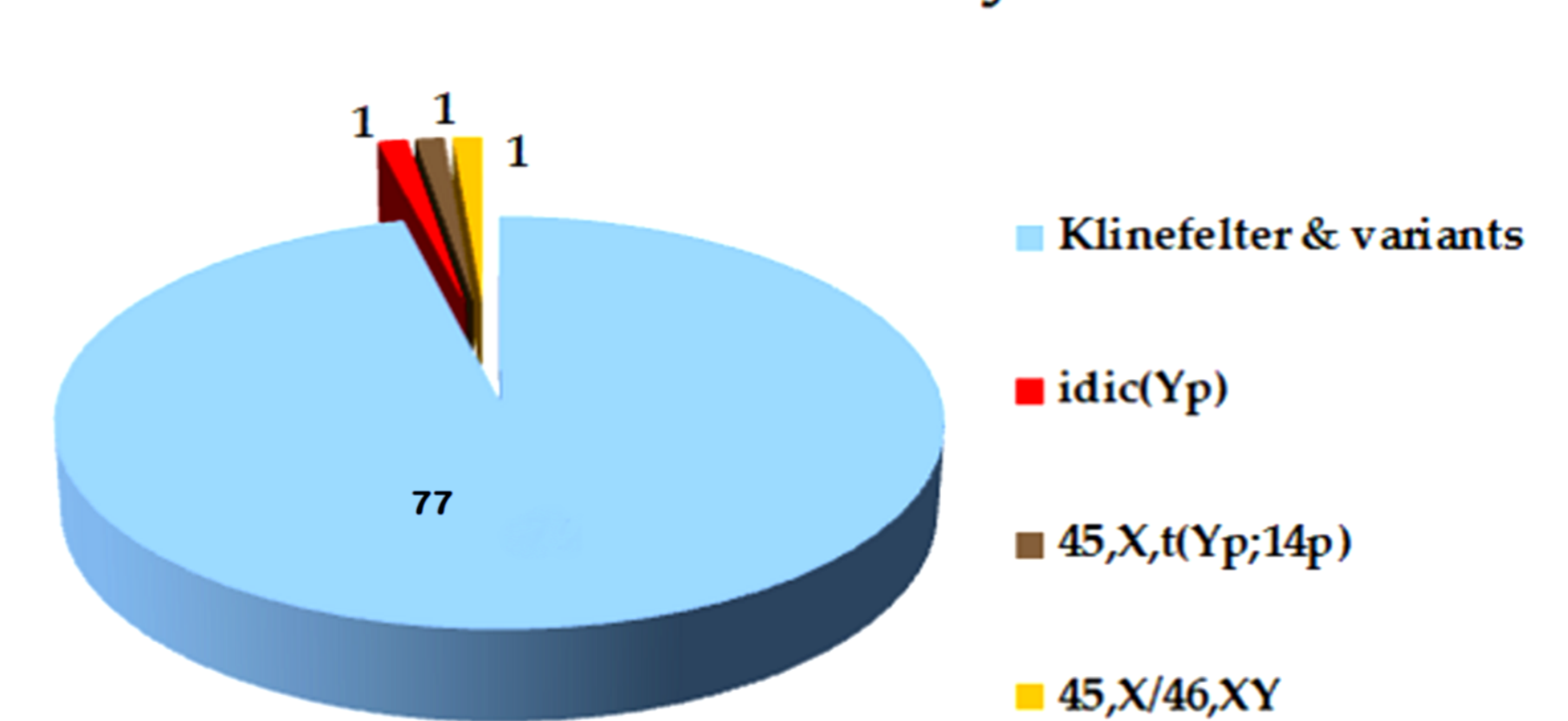
Primary amenorrhea/ female infertility



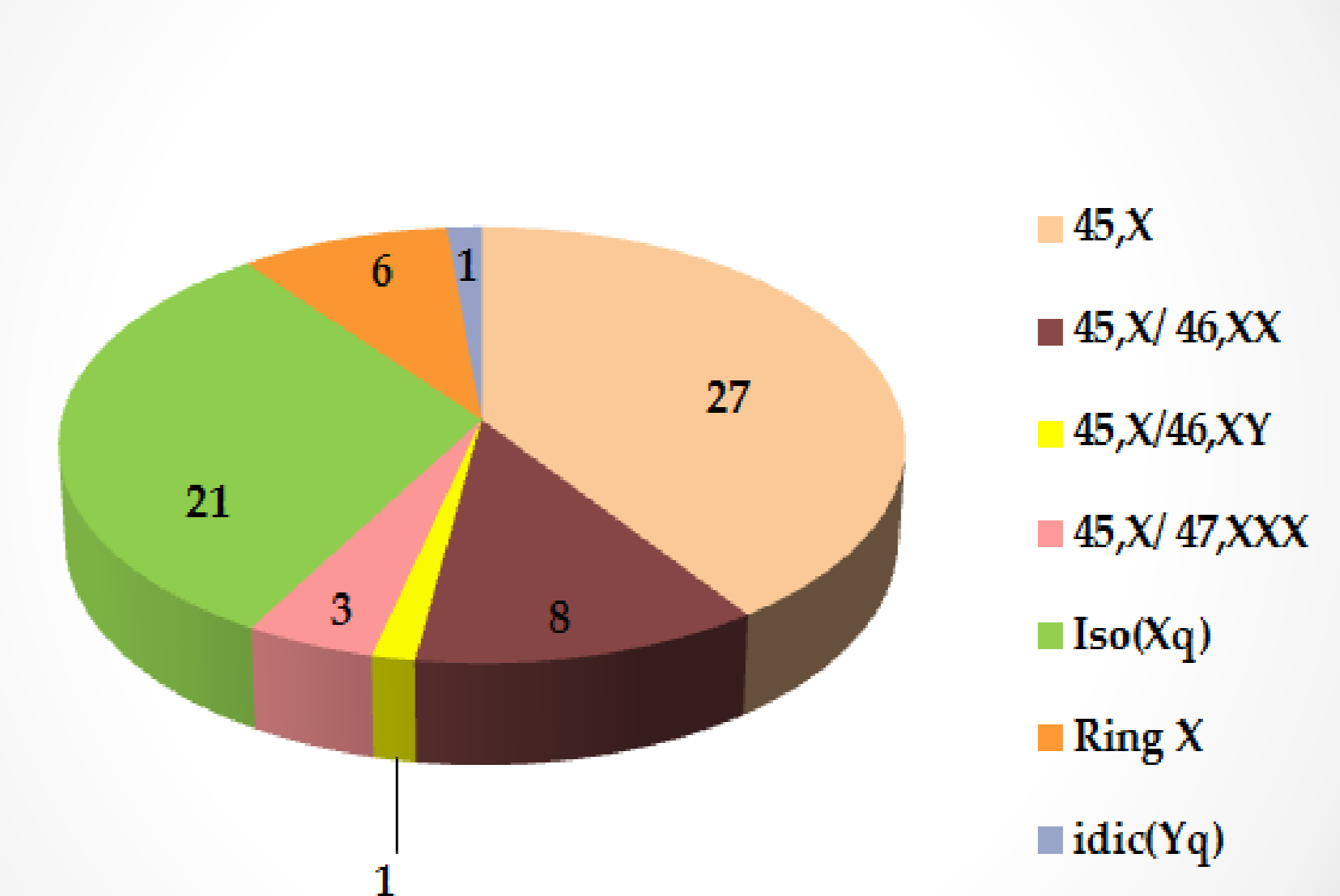
Karyotype and FISH analysis of a patient with ambiguous genitalia and isodicentric (Yq) abnormality.



Male Infertility



Short stature, TS manifestations



## CONCLUSIONS

- Sex chromosomal abnormalities constitute a high proportion of DSD
- Application of ACGH is necessary for diagnosing MCA with DSD
- Detection of Y abnormalities in MGD is necessary for genetic counseling and gonadectomy (dysgerminoma)

