# A new interpretation 46,XX karyotype in male after localization SRY gene using Fluorescence *In Situ* Hybridization

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## Introduction

46,XX Disorder of Sexual Development (DSD) is a rare condition associated with male infertility in all cases. The next necessary examination after 46,XX karyotype detection in male is the diagnostics of SRY+/- syndrome variants. Method FISH (Fluorescence In Situ Hybridization) has some benefits in comparison with molecular diagnostics – not only detection SRY presence, but its localization in the karyotype and mosaicism identification. The aim of the study was to determine gene SRY location in karyotype. Methods: GTG-banding (550 bands, 25 metaphases), FISH.

## Results

SRY localization was analysed in 11 46,XX male patients by FISH: 7 adults with azoospermia, 1 oligozoospermia, 3 boys with suspected Klinfelter syndrome (KS) and congenital developmental features (Figure 1). In addition to the most common well-known SRY+variant der(X)t(X;Y)(p22.3;p11.2) gene SRY was detected in 3 karyotypes too. In all these cases, after the initial stage of hybridization with the LSI SRY probe, subsequent karyotype reanalysis and hybridization rounds with appropriate DNA probes were carried out according to the established abnormality.

Patient 1: Initially SRY location was detected at the short arm one of D-group chromosomes in 28 years old male with azoospermia. Der14 as a result cryptic t(Y;14) in parental meiosis was detected after karyotype reanalysis and using 14 chromosome LSI probe (Figure 2).

Patient 2: Presence of two Y-chromosomes low level mosaicism (1% cells) are associated with oligozoospermia in 25 years old male and so gives a chance to natural fatherhood.

Patient 3: KS with low level mosaic clone abnormal Y-chromosome psuidic(Y)(q11.2) (DXYS129/153++,SRY++,DYZ3++) was confirmed in a 14 years old boy (Figure 3).

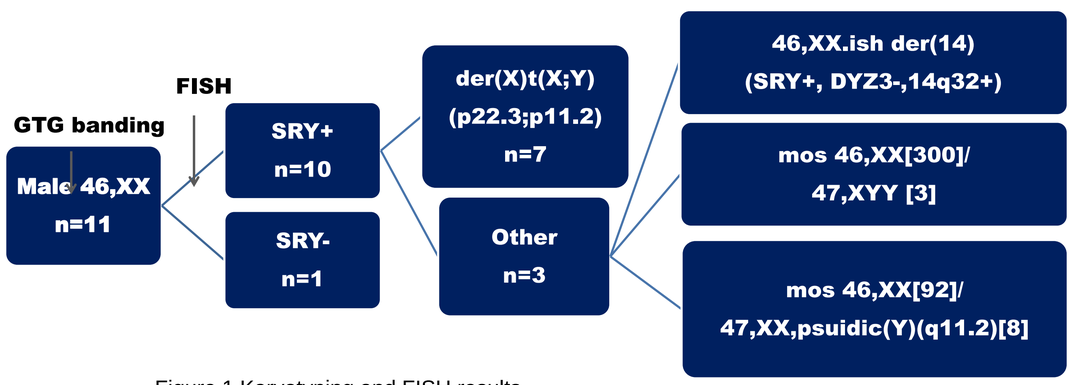


Figure 1 Karyotyping and FISH results

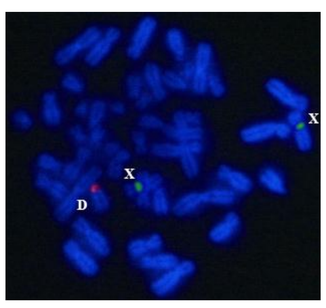


Figure 2a Metaphase FISH/46,XX,der(14)(SRY+,14q32+): LSI SRY SpectrumOrange/ CEPX(DXZ1) SpectrumGreen (Abb.Mol.)

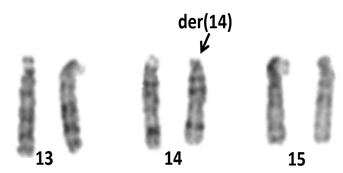


Figure 2b Metaphase GTG banding/ 46,XX,der(14)(SRY+,14q32+): D-group chromosomes

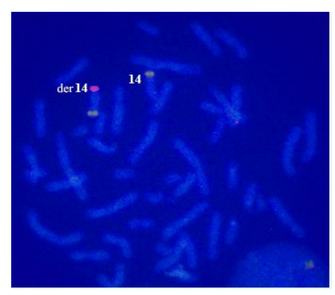


Figure 2c Metaphase FISH/ 46,XX,der(14)(SRY+,14q32+): LSI SRYSpectrumOrange, LSI IGH Dual SpectrumOrange+Green (14q32)Color(Abb.Mol.)

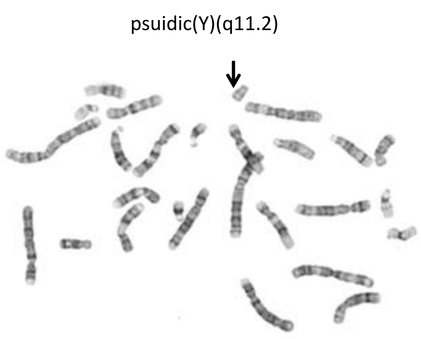


Figure 3a Metaphase chromosomes GTG banding/mos45,X/46,X,psuidic(Y)(q11.2)

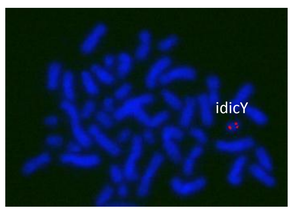


Figure 3b Metaphase FISH/ mos45,X/46,X,psuidic(Y)(q11.2): DYZ3 Spectrum Orange (Abb.Mol.)

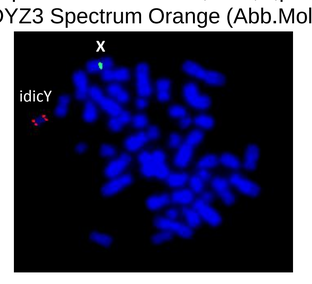


Figure 3c Metaphase FISH/ mos45,X/46,X,psuidic(Y)(q11.2): LSI SRY SpectrumOrange/DXZ1 Spectrum Green (Abb.Mol.)

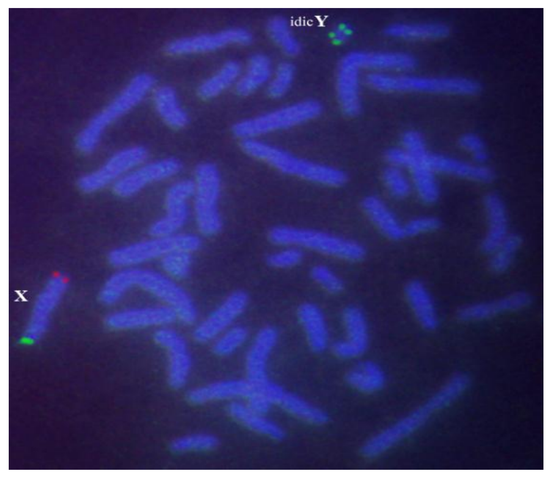


Figure 3d Metaphase FISH/ mos45,X/46,X,psuidic(Y)(q11.2): TelVysion Xp/Yp SpectrumGreen, TelVysion Xq/Yq SpectrumOrange (Abb.Mol.)

## Conclusion

Clarification of the karyotype is essential for the correct medical genetic counseling. Our study has pointed out the importance of FISH in the correct determination of the karyotype, the subsequent analysis of genotype-phenotype correlations, psychological support, genetic counseling of these patients.

## References

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