A rare case of infertility – SRY positive, 46XX testicular disorder of sexual differentiation

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BACKGROUND

The testicular disorder of sexual differentiation (DSD) is a rare clinical condition with a reported incidence of 1:20,000 newborn males. It is characterized by a male phenotype with a 46XX karyotype. There are three clinical phenotypes: normal males, males with genital ambiguity and males who are true hermaphrodites. SRY positive 46XX testicular DSD results from the translocation of a Y chromosome segment containing the SRY gene during spermatogenesis. In rare cases, the SRY gene may be misplaced onto a chromosome other than the X chromosome.

CASE REPORT

A 33 year old male presented with his wife to the fertility clinic at Galway University Hospital with a three year history of primary infertility.
The patient's wife had no significant past medical history. Her clinical examination was unremarkable and her biochemical and hormonal investigations were normal.
The patient had a past medical history of appendectomy, lumbar discopathy and undescended testes in childhood. His family history was not significant. He reported normal libido and sexual function. Clinical examination revealed him to have normal adult male height, normal secondary sexual characteristics and bilateral small testes.
Male investigations included a sperm sample which revealed azoospermia. The patient had chromosomal analysis performed using fluorescence in situ hybridization (FISH) technique. This confirmed a 46XX SRY positive karyotype with translocation of the SRY gene between the X and the Y chromosome – 46XX der(X)t(X;Y)(p22.3;p11.3)(SRY+).

BLOOD RESULTS

Blood Test	Value
LH	18.7 IU/l
FSH	40.1 IU/l
Testosterone	5.2 nmol/l
Viral screen	Negative
Cystic Fibrosis gene	Negative
Vitamin D	48 nmol/l

DXA SCAN

Z score L1-L4	**-1.2**
Z score NOF	**-1.2**

MRI PELVIS

No female organs were visualised. The seminal vesicles were not distended.
Conclusion: an unremarkable MRI of male pelvis.

TREATMENT

The patient was commenced on testosterone therapy and vitamin D replacement. The couple underwent fertility treatment using donor sperm which resulted in a successful pregnancy.

CONCLUSIONS

Patients with azoospermia should have chromosomal analysis performed. Sperm donation remains a fertility treatment option for patients with DSD and had a successful outcome in this patient.
Patients with DSD require lifelong follow-up led by an endocrinologist. These patients should have testosterone supplementation, regular imaging of gonads, interval bone density measurements, and access to psychological support and fertility treatment as required.

References

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ECE 2016 327-EP
Clinical case reports - Thyroid/Others
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Poster presented at: ECE2016 28 – 31 May 2016 MUNICH, GERMANY