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A Case Report on Klinefelter's Syndrome

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Abstract

Azoospermia is present in 1% of all men and approximately 15% of infertile men. Incidence of Klinefelter's syndrome is 1 in 500. Mr. X, a 38 years old motor cycle driver presented with postcoital scrotal pain along with a recent semen analysis report showing azoospermia. O/E, bilateral small testes (no H/O mumps orchitis) found & repeat semen analysis after 6 weeks revealed azoospermia in centrifuged semen. Raised S.FSH (29.41 mU/ml) & S. LH (24.40mU/ml), low S. Testosterone (6.04 nmol/L) and normal S. Prolactin level. He was provisionally diagnosed as hyper gonadotrophic hypogonadism or primary testicular failure. Then Karyotyping showed Klinefelter's syndrome (47XXY). Counselling was done on PESA/TESE and scope of ICSI, but due to financial constrain these procedures could not be done. Both patient & his wife have been counselled regarding donor sperm IUI &/or adoption.

Abstract: Azoospermia, Karyotype, Klinefelter's Syndrome

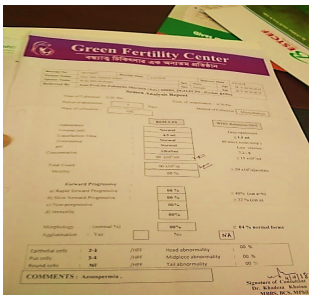
Introduction

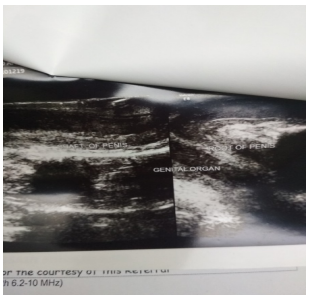
Azoospermia is present in 1% of all men[1] and approximately 15% of infertile men[2]. Incidence of Klinefelter's syndrome is 1 in 500. It includes 1% of all men attending infertility clinic. It is associated with 47XXY karyotype in nonmosaic forms & 46XY/ 47XXY in mosaic forms.

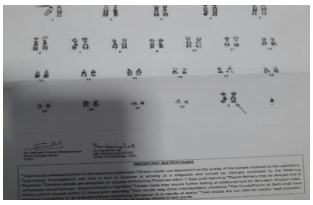
Case report

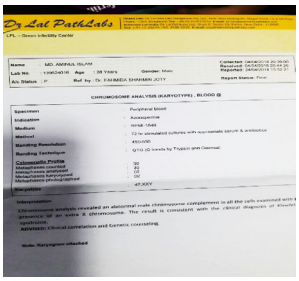
Mr. X, a 38 years old motor cycle driver presented with postcoital scrotal pain along with a recent semen analysis report showing azoospermia. He was married for 8 years without issue & his wife left for this. He is married again for last 6 years without any offspring. All clinical, laboratory & radiological findings of her wife is with in normal limit. The patient was referred to urologist & examination revealed bilateral small testes (no H/O mumps orchitis). Repeat semen analysis was advised after 6 weeks which revealed azoospermia in centrifuged semen.

USG of genital organ revealed bilateral small testes (Rt 18x5mm, Lt 17x7mm). Hormone analysis revealed raised S.FSH (29.41 mU/ml) and S. LH (24.40mU/ml), low S. Testosterone (6.04 nmol/L) and normal S. Prolactin level. He was provisionally diagnosed as hyper gonadotrophic hypogonadism or primary testicular failure.
Then Karyotyping showed Klinefelter's syndrome (47XXY). Counselling was done on PESA/TESE and scope of ICSI, but due to financial constrain these procedures could not be done. Both patient & his wife have been counselled regarding donor sperm IUI &/or adoption.









Discussion

Azoospermia can be diagnosed when no spermatozoa is detected on high-powered microscopic examination of centrifuged seminal fluid (3000xg for 15 min) on at least 2 occassions[3]. It can be either pre testicular/ testicular/ post testicular, or obstructive/ nonobstructive. To evaluate an azoospermic male detailed medical history about prior fertility, viral orchitis/ cryptorchidism, genital trauma/ surgery/ infection, prior RT/CT, recent fever, heat exposure & current medication should be taken. F/H/O birth defects, mental retardation, reproductive failure or cystic fibrosis should be evaluated. Examination of testicular, vas & epididymal size/ consistency, secondary

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sex character (body hair distribution, gynaecomastia), varicoceles & DRE for masses should be done. If S. FSH & S. testosterone is normal then trans rectal USG & vasography should be done to evaluate reproductive tract obstruction & testicular biopsy can be taken. If normal spermatogenesis was seen then obstructive azoospermia; if abnormal, then hypospermatogenesis/ maturation arrest. If low S. FSH & S. testosterone then hypogonadotrophic hypogonadism/ pretesticular azoospermia & if high S. FSH, low S. testosterone then primary testicular failure or hypergonadotrophic hypogonadism / non obstructive azoospermia. Finally, genetic testing (Karyotyping/ Y chromosome micro deletion) should be done to detect the cause of primary testicular failure. Proper counselling regarding surgery/ ICSI/ adoption/ Donor sperm IUI should be given according to cause.

Klinefelter's syndrome is the most common chromosomal abnormality associated with primary testicular disease or non obstructive azoospermia. In mosaic variety, individuals can rarely present with severe oligospermia & natural pregnancies, but this gradually declines with age. On clinical examination, signs of androgen deficiency (due to high S. FSH & low S. testosterone) are seen with small & firm to hard testes with loss of libido & sexual dysfunction[4]. Chances of sperm retrieval after microdissection TESE in Klinefelter's syndrome varies between 30-45%[5].

Conclusion

With the advent of ICSI, a new ray of hope has emerged as a possible chance to attain paternity in azoospermia6. Healthy sperm if retrieved from klinefelter testes will be of chromosomal competence in vast majority & can give birth to healthy babies.

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