Should cryopreservation of sperm be routinely offered to Klinefelter’s patients?

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Case Report

A couple were referred to the unit as the male partner had been diagnosed with non-mosaic Klinefelter’s Syndrome (47XXY). An IVF & ICSI cycle was embarked upon and the male partner underwent TESE (Schlegel et al. 1997). Post preparation of the testicular biopsies, all 7 oocytes collected were inseminated using the ICSI procedure 41 hours post hCG (Palermo et al. 1992). At 18hpi, six oocytes showed signs of normal fertilisation (2PN and 2PB). On day five of development two embryos were transferred to the uterus resulting in a live birth at 39 weeks + 5 days with a normal karyotype.

Despite published evidence of healthy live births from Klinefelter’s patients using ICSI with testicular or ejaculate sperm, it is currently not common practice in the UK to offer surgical sperm retrieval and cryopreservation as an option for fertility preservation in these men (Fullerton et al. 2010).

One of the main oppositions to fertility treatment for patients with Klinefelter’s Syndrome is the potential genetic risks to the offspring.

Although the risk of sex chromosome and autosomal aneuploidy has shown to be increased in comparison to the general population, this increase is comparable to that seen in surgically retrieved sperm from azoospermic men (range of 1.5% to 11.4%) with a normal karyotype (Levron et al. 2000; Palermo et al. 2002).

A similar increased frequency of autosomal aneuploidy (ranging from 0-10%) has also been demonstrated in 46XY males with oligozoospermia or oligoasthenoteratozoospermia (Pfeffer et al. 1999; Morel et al. 2003).

Both groups of patients are referred for ICSI treatment routinely, which questions why Klinefelter’s patients are not commonly advised to undergo cryopreservation for fertility preservation.

References