

Disorder of sexual development in a male pup with inverted 'Y' chromosome

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Two German Shepherd puppies were delivered by Caesarean section following a pregnancy conceived from a dual sire fresh and frozen semen insemination. On assessment at delivery one pup was a phenotypic female and the other was ambiguous with an external urethral opening approximately halfway between the normal position of the male just caudal to the umbilicus and a normal female. This was consistent with a hypospadias. At 18 days of age blood samples were collected from both pups to determine parentage and karyotype. Both pups were conceived from the frozen semen insemination. Karyotyping of the phenotypically normal female was consistent with 78 XX. However, the ambiguous pup demonstrated a male karyotype with two related cell lines. One line exhibited a normally structured Y chromosome. In the other cell line the Y chromosome appeared to be inverted resulting in an acrocentric structure and not metacentric as in the normal Y chromosome. It is probable that the observed cytogenetic alteration interfered with a balance and expression of genes in the Y chromosome leading to a developmental aberration. Reassessment of this pup at 88 days of age revealed no functional urinary deficits but no palpably descended testicles consistent with bilateral cryptorchidism. This dog is classified as a 78XY/78X invertedY chimaera. It has not undergone gonadectomy so microscopic assessments of his gonads has not been performed.