Newborn whit a derivative chromosome X and ambiguous genitalia

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Translocations involving the short arms of the X and Y in human chromosomes are uncommon. One of the primary functions of the X and Y chromosomes is gender phenotype determination.
Here we report a newborn female with ambiguous genitalia and abnormal X chromosome.
Karyotype was performed using the standard methods and Fluorescence *in situ* hybridization (FISH) directed for the *SRY* gene was used for confirmation of the clinical and cytogenetic suspicion. Chromosomal *microarray* analysis (CMA) was performed using CytoScan HD (Affimetrix®) to identified gains/loses on the der(X) chromosome.
The analyse revealed one abnormal X chromosome in a female karyotype. Considering the ambiguous genitalia clinical information the abnormal X was considered to be compatible with a translocation X/Y. This was confirmed by the presence of signal for the *SRY* using FISH.
CMA allowed to clarify a loss of 12.34 Mb at Xp22.33p22.2 and a gain of 7.41 Mb at Yp11.31p11.2 (ISCN = arr[GRCh37] Xp22.33p22.2(2703632_15050955)x1,Yp11.31p11.2(2650140_10059230)x1).
The X deleted region includes several OMIM morbid genes, including *CLCN4*. Mutations in *CLCN4* are associated with intellectual disability and impaired language development, and heterozygous females can be as severely affected as male. The gain on the Y encompasses nine OMIM genes, including the *SRY* gene, involved in the sexual male development. This additional information can be of great value for the child development.
Translocations of segments of Y chromosome containing *SRY* are described in sexual reversion and true hermafroditism cases, which could explain the reason for referral for the newborn.
Nevertheless, translocations between the X/Y chromosomes in females are expected to have a skewed inativation pattern in favour of the abnormal X and X-inativation studies could prove this likelihood. If a normal developmental of the child is observed over time this will be likely due to the preferable inativation of the abnormal X.
Presently the child is about 1-year-old and she presents normal uterus, ovarian, and external genitalia, with absence of male gonads. No other clinical features have been identified.