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A cytogenetic investigation on a suspected pseudo-hermaphroditism clinical case in "Rhoen-sheep"

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A lamb of the German "Rhoen-sheep" breed was born spontaneously with deformities of genital organs (pseudovagina with labioscrotal beadings) and urethra after normal twin pregnancy. Its twin brother was healthy and showed no deformities. Classical and molecular cytogenetic investigations were carried out to study possible karyotype defects responsible for the abnormal phenotype. Peripheral blood sample cultures were performed to get both normal and BrdU-treated cultures, the latter to obtain R-banded preparations. Normal cultures were used to perform CBA-banding and FISH-technique. The analysis of the C-banding proved the correct position of the centromeres, whereas the RBA-banding pattern showed karyologically normal arrangement (2n=54,XY). A FISH analysis was carried out to evaluate the eventual level of XX/XY mosaicism by using specie-specific painting probes for sex chromosomes. 100 metaphases were scored and all showed normal XY chromosomal arrangement. No metaphases with two X chromosomes were detected. The observed phenotype and the lack of cytogenetic defects led to state that this clinical case might represent a suspected condition of male pseudo-hermaphroditism. In humans, this condition is related to the androgen insensitivity syndrome (AIS). Further investigation is therefore necessary to identify at molecular level the causes of this abnormal phenotype.