

AperTO - Archivio Istituzionale Open Access dell'Università di Torino

**A cytogenetic investigation on a suspected pseudo-hermaphroditism clinical case in “Rhoen-sheep”.**

**This is the author's manuscript**

*Original Citation:*

*Availability:*

This version is available <http://hdl.handle.net/2318/150206> since

*Published version:*

DOI:10.1007/s10577-014-9435-7

*Terms of use:*

Open Access

Anyone can freely access the full text of works made available as "Open Access". Works made available under a Creative Commons license can be used according to the terms and conditions of said license. Use of all other works requires consent of the right holder (author or publisher) if not exempted from copyright protection by the applicable law.

(Article begins on next page)



UNIVERSITÀ DEGLI STUDI DI TORINO

*The final publication is available at Springer via <http://dx.doi.org/10.1007/s10577-014-9435-7>*

**P8**

**A cytogenetic investigation on a suspected pseudo-hermaphroditism clinical case in “Rhoen-sheep”**

A. Pauciullo<sup>1,2</sup>, D. Schiefen<sup>1</sup>, G. Lühken<sup>1</sup>, G. Erhardt<sup>1</sup> ([alfredo.pauciullo@agrar.uni-giessen.de](mailto:alfredo.pauciullo@agrar.uni-giessen.de))

<sup>1</sup>Justus Liebig University, Institute for Animal Breeding and Genetics, Giessen, Germany;

<sup>2</sup>National Research Council (CNR), ISPAAM, Laboratory of Animal Cytogenetics and Gene Mapping, Naples, Italy

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.