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A cytogenetic investigation on the Lethal White Syndrome in sheep

Pauciullo A.^{1,*}, Fett K.¹, Lühken G.¹, Parma P.², Di Bernardino D.³, Erhardt G.¹

¹Institut für Tierzucht und Haustiergenetik, Justus-Liebig-Universität, Gießen, Deutschland

²Dipartimento di Produzione Animale, Università di Milano, Milano, Italia

³Dipartimento di Scienza del Suolo, della Pianta, dell'Ambiente e delle Produzioni Animali, Università di Napoli "Federico II", Portici (NA), Italia

*Corresponding author: alfredo.pauciullo@agr.uni-giessen.de

The mating of genetically related animals is known to increase the homozygosity. Such condition can also increase the chances of the offsprings to be affected by recessive traits. Lethal white syndrome (LWS), is an autosomal genetic disorder most prevalent in the American Paint Horse. Affected animals phenotypically appear normal, although they have all-white coats and blue eyes. Internally, they have a non-functioning colon and die within a few days after birth.

In a small group of Cameroon sheep, where only one ram was used for several consecutive years and mated to his relatives, five lambs were completely white-coated with blue eyes. All died shortly after birth. A cytogenetic investigation was carried out on the available mothers, most probably heterozygous carriers of the genetic defect. Peripheral blood sample cultures were performed for two ewes 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, while R-banded preparations were used for both karyotyping (RBA-banding) and RBPI-FISH technique.

The analysis of the CBA- and RBA-banding pattern showed karyologically normal arrangement ($2n=54,XX$). LWS syndrome has been compared to Hirschsprung's disease in humans, which is caused by mutations on the EDNRB gene. A conventional FISH analysis carried out by using a BAC probe, showed EDNRB gene deletion at heterozygous status. A RBPI-FISH experiment was additionally performed to clarify the position of the gene along the chromosome. According to standard ideogram, the EDNRB gene was confirmed to be located on the OAR10q2.2. This finding confirmed that the ewes were heterozygous carriers of EDNRB gene deletion.