MOLECULAR DIAGNOSIS OF INHERITED DISEASES

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For several years, agricultural production has been affected economically because of many inherited diseases described worldwide. According to recent bibliography there are 357 diseases and traits produced by a single locus, with 53 caused by identified mutations by molecular diagnosis. Many of these diseases are scattered through the world because of the importation of semen. The intensive breeding of sires having inherited diseases predisposes a wide and rapid dissemination producing perinatal mortality. Our country has made molecular diagnosis for several of these diseases in institutions like INIA, DILAVE or FV. Recently, Uruguay has approved the amendment of Law 18 341 (article 14) that encourages the verification of semen, embryos and breeding against these diseases and encourages the usage of new techniques of diagnose. The aim of our work is to develop molecular diagnostic techniques that allow identify diseases before they could be used in reproduction. We develop PCR-RFLP techniques for molecular diagnosis for the following diseases in beef cattle: MSUD (Maple Syrup Urine Disease), Epidermolysis Bullosa and α-Manosidosis and for dairy cattle: BLAD (Bovine Leucocyte Adhesion Deficiency) and Citrulinemia. We used blood and tissue samples sent by DILAVE. MSUD generates fatal neurological symptoms in the first days of life. We successfully diagnose MSUD 248 mutation in 5 animals. Epidermolysis Bullosa causes separation of the dermo-epidermal junction by a keratin 5 gene mutation. After studies, 3 animals were detected as carriers of the disease. The α-Mannosidosis causes a fatal nervous symptom. Until now we have diagnosed the disease in 3 samples out of 31 analyzed. BLAD (Bovine Leukocyte Adhesion Deficiency) is a disease characterized by recurrent bacterial infections in calves 8-9 months of age by reduced maternal immunity, due to the mutation of a membrane glycoprotein (CD18) of leukocytes. We analyzed 251 cows in dairy herd animals only resulting two carriers. The Citrulinemia causes a lethal disease in newborn calves generated by the mutation of the enzyme ASS, but studies for the animal were not detected as carriers of the disease.

We conclude that the molecular diagnosis let detect in our country 4 inherited diseases of 5 diseases studied.