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Innovative therapies for inherited skin disorders

Monogenic skin diseases, also known as genodermatoses, constitute about 7-8% of rare diseases and together represent a significant part of dermatology. Currently the genetic basis of more than 400 genodermatoses, have been elucidated which allows classification, molecular diagnosis, understanding their pathogenic mechanisms and finally the development of safe and effective curative evidence-based therapies treatments. As with other types of rare diseases, often the consequences of genodermatosis are devastating for the patients, their families and friends.

Genetic skin diseases comprise epithelial adhesion, keratinization, pigmentation, DNA repair, and connective tissue disorders. In this conference, the focus will be set on epithelial adhesion disorders, ie epidermolysis bullosa (EB). EB comprises a group of skin diseases characterized by defects in epithelial-mesenchymal adhesion. EB is genetically and clinically heterogeneous. Phenotypes range from mild to severe muco-cutaneous presentations. In the last years, remarkable advances in the development of innovative treatments for EB have been achieved. The strategies include, read-through drugs, cell therapy, tissue bioengineering and gene therapy approaches (viral and non-viral) employing exon skipping, gene addition and gene editing strategies among others. During the conference preclinical and clinical breakthrough from our laboratory and others will be thoroughly reviewed and discussed.

Biography

Marcela Del Rio Nechaevsky is full Professor of Experimental Dermatology at the University Carlos III de Madrid (Spain). She is the head of the Unit 704 at Centre for Biomedical Research on Rare Disease (CIBERER), as well as the director of the Regenerative Medicine and Bioengineering Group at the Fundación Jimenez Diaz hospital. She has worked on regenerative medicine for more than 20 years. Her current research interest lies in discovering what causes inherited skin diseases and what can be done to develop new clinically relevant, evidence-based personalized medicine for people living with inherited disorders of the skin. She now leads and collaborates on several National and International projects to develop advanced therapies (cell-, gene- and tissue engineered based therapies).

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