GENETICS AND GENODERMATOSES

# RARE GENODERMATOSES IN OUR DERMATOLOGIC PRACTICE

*Hana Zelenkova, M.d., Ph.d., Mba* [(1)]

*Sanare Spol. S. R.o., Work Place Private Clinic Of Dermatovenerology, Dost Svidník, Svidník, Slovak Republic* [(1)]

Background: The paper refers to rare genodermatoses observed and treated in our dermatologic practice.

Observation: Tricho-dento-osseous (TDO) syndrome is an autosomal dominant genetic disorder that belongs to a group of diseases known as ectodermal dysplasias. Ectodermal dysplasia typically affects the hair, teeth, nails, and/or skin.
Asymmetrically localised naevi flammei may be associated with other conditions such as Sturger-Weber syndrome and Klippel-Trenaunay syndrome.
Melanosis neurocutanea Touraine (1941), Neurocutaneous melanosis, (MNC), originally described by Rokitansky (1861), is considered to be a sporadic congenital disorder, a non-hereditary melanphacomatosis (incidence in 1/20000 newborns). The syndrome affects both genders alike (it has until now been only described in Caucasians), and is characterised by very large or multiple pigmented cutaneous nevi as well as hyperploriferation of benign and malignant melanin cells in the leptomeninges.
Ichtyosis – Collodion baby. Heterogeneous group of diseases. The disorder affects the entire skin. Divided based on examination and clinical, genetic, histological, ultra-structural and sometimes biochemical signs
Acrodermatitis enteropatica - Autosomal recessive metabolic disorder influencing zinc uptake
The uncombable hair syndrome is also known as Einstein Syndrome, although Albert Einstein himself was not a sufferer. The syndrome is caused by congenital genetic mutation that influences the colour and structure as well as the bizarre appearance of hair. It is causes by the mutation of one of the following three genes: PADI3, TGM3, and TCHH.

Key words: rare genodermatoses