ABSTRACT BOOK ABSTRACTS



A new ERA for global Dermatology 10 - 15 JUNE 2019 MILAN, ITALY

GENETICS AND GENODERMATOSES

HYPOHIDROTIC ECTODERMAL DYSPLASIA (HED): REPORT OF 24 NEW CASES

A. $Veleda^{(1)} - M$. Fiandrino⁽²⁾ - N. Primc⁽²⁾ - R. Alperovich⁽²⁾ - M. E. Abad⁽³⁾ - M. Boente⁽²⁾ - M. Larralde⁽³⁾

Hospital Ramos Mejía, Dermatología Infantil, Buenos Aires, Argentina⁽¹⁾ - Hospital El Niño Jesús, Centro Integral De Dermatología, Tucumán, Argentina⁽²⁾ - Hospital Ramos Mejía, Hospital Alemán, Dermatología, Buenos Aires, Argentina⁽³⁾

Background: Hypohidrotic ectodermal dysplasia (HED) is a congenital condition defined by reduced or absent development of teeth, hair follicles and/or eccrine sweat glands. It is caused by mutation of the genes encoding ectodysplasin A (EDA), EDA receptor (EDAR), or its intracellular adapter protein EDARADD. Recently intraamniotically recombinant protein-replacement therapy (receptor-binding domain of EDA) allowed adequate development of sweat glands.

Observation: We report 24 new cases of HED (17 males and 7 females) from 3 different centres seen in the last 14 years. Most cases had an affected family member. Two were born as collodion babies. Characteristic dysmorphic facial features (frontal bossing, low nasal bridge, palpebral hyperpigmentation, flat malar ridges, everted thick lips) were seen in all of them. Hypotricosis with dull hair was also a common feature. Dryness of skin, eczema and ichthyosiform skin was seen in all the cases. The female patients presented less conspicuous features; mosaic hyperpigmentation or mosaic hair anomalies were seen in 3 of the 7 women. Hyperthermia was a common finding. Otitis media that lead to chronic sinus infections was present in one patient who developed a maxillary sinus mucocele.

Key Message: We report these cases in order to highlight the already known features of HED. The frequent familial cases diagnosed in this series and the less conspicuous signs in females point towards the need to investigate women in the family. Promising treatment during pregnancy needs further investigations with larger number of patients.



24TH WORLD CONGRESS OF DERMATOLOGY MILAN 2019



International League of Dermatological Societies *Skin Health for the World*

