

RUDOLF HAPPLE AND THE DERMATOLOGY EPONYMS LINKED TO HIS NAME

RUDOLF HAPPLE I DERMATOLOGICZNE EPONIMY ZWIĄZANE Z JEGO IMIENIEM

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Abstract

Rudolf Happle is a well-known pediatric dermatologist. He reported and described several medical conditions and syndromes. Some of these are linked eponymously to his name.

This concise letter sheds light on eponymic designations which bear his name.

Streszczenie

Rudolf Happle jest znanym dermatologiem dziecięcym. Przedstawił i opisał kilka stanów medycznych i zespołów chorobowych. Niektóre z nich są związane z jego imieniem.

Ten zwięzły list rzuca światło na nazwy jednostek chorobowych noszących jego imię.

Key words: dermatology; eponym; syndrome

Słowa kluczowe: dermatologia; eponim; zespół chorobowy

Professor Rudolf Happle (Fig.1), is a world-renowned dermatologist, with great contributions to dermatology in general and pediatric dermatology in particular. Professor Happle was born in 1938 in Freiburg, Germany [1]. He is currently, a Professor Emeritus of Dermatology in the Department of Dermatology at the University of Marburg in Germany and a Professor Emeritus at the Department of Dermatology, Philipp University. After his retirement, he is working as a Guest Professor in the Department of Dermatology at the University of Freiburg, Germany.

Dr Happle reported and described several dermatological conditions and syndromes. Some of these are linked eponymously to his name. The following paragraphs highlight some eponyms linked to his name.

Conradi-Hünemann-Happle syndrome (CHHS)

CHHS is (MIM#302960) is a X-chromosomal dominant disorder that usually affects only females and is lethal in males. It has cutaneous, skeletal, and ocular manifestations; it also is referred to as X-linked dominant chondrodysplasia punctata or Happle syndrome. It was fully delineated by Happle between 1977 and 1981 as an X-linked gene defect. The clinical phenotype of the CHH syndrome is variable,

ranging from stillborn or lethal forms to mild, clinically almost undetectable forms. The clinical hallmarks of the CHH syndrome are linear ichthyosis, chondrodysplasia punctata, asymmetrically shortened limbs, unilateral, and usually sectorial, cataracts, and short stature [2].

Patients with CHHS are born with ichthyosiform erythroderma that is characterized by feathery, adherent hyperkeratosis and a distribution along Blaschko lines.

The erythroderma usually resolves spontaneously during the first months of life. Subsequently, residual streaks and swirls of follicular atrophoderma and, occasionally, hyper- or hypopigmentation are noted. Scalp involvement results in patchy, scarring alopecia. Skeletal abnormalities include short stature, craniofacial anomalies, asymmetric limb reduction defects, vertebral malformations, and hip dysplasia. The sign of stippled calcifications of the epiphyses (chondrodysplasia punctata) [2] can be noted on x-rays only during the first months of life.

CHHS is caused by mutations in the gene that encodes the emopamil binding protein (EBP), causing a defect in sterol biosynthesis pathway. The EBP gene resides on the short arm of the X chromosome.

Ruggieri-Happle syndrome

This is a particular type of cutis tricolor (combination of congenital hyper- and hypopigmented skin lesions in close proximity to each other on a background of

normal complexion), when it occurs as a part of a complex malformation syndrome.

Cutis tricolor may be a marker of underlying skeletal or neurological involvement [3].



Figure 1. Rudolf Happle with the author during the 10th International Congress of Dermatology, Prague, Czech Republic, May 20-24, 2009

Happle-Tinschert syndrome

Happle and Tinschert described the case of a multisystem birth defect with segmentally arranged basaloid follicular hamartomas associated with extracutaneous defects in the form of short leg, polydactyly and hypoplastic teeth. They presented a comprehensive overview of 8 similar cases reported under various designations, and provided evidence that this syndrome includes various additional defects of the bones, teeth and brain. This syndrome was later named Happle-Tinschert syndrome [4].

Other conditions

It is not uncommon to find the name of Professor Happle in the titles of other dermatological conditions to which he contributed by his great researches. For example the types of segmental involvement in autosomal dominant diseases [5]. Also, in phacomatosis pigmentokeratocica, which is a rare and distinct variant of the epidermal nevus syndrome, first described by Happle et al. It comprises the association of a linear organoid (epidermal) nevus with sebaceous differentiation and a speckled lentiginous nevus of the papular type arranged in a checkerboard pattern [6].

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