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JADWIGA SCHWANN AND HER SYNDROME

JADWIGA SCHWANN I OPISANY PRZEZ NIĄ ZESPÓŁ CHOROBOWY

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Abstract
Jadwiga Schwann was a dermatologist from Poland. In the 1960s, Schwann reported a rare congenital genodermatosis. This syndrome is characterized by knuckle pads, leukonychia, palmoplanter keratoderma and sensorineural deafness. This report sheds light on Schwann and the syndrome that bears her name.

Streszczenie
Jadwiga Schwann była dermatologiem z Polski. W 1960 roku Schwann odnotowała rzadką, wrodzoną genodermatozę. Zespół ten charakteryzuje się objawem knuckle pads, leukonychią, rogowcem dłoni i stóp, głuchotą czuciowo-nerwową. Raport ten rzuca światło na J. Schwann i zespół objawów, który nosi jej imię.

Key words: Jadwiga Schwann; congenital genodermatosis; Bart - Pumphrey syndrome
Słowa klucze: Jadwiga Schwann; congenital genodermatosis; Bart - Pumphrey syndrome

Jadwiga Schwann was a dermatologist from Poland. Among her contributions to dermatology, she is credited for describing a syndrome, in German and Polish languagues [1,2]. This syndrome appeared latter in English literature by Robert S. Bart (Dermatologist) and Robert E. Pumphrey (Otolaryngologist) [3]; both from USA, and so the syndrome was then known as Bart - Pumphrey syndrome [4-10].

Schwann syndrome is cited in the Online Mendelian Inheritance in Man [10], as knuckle pads, leukonychia, and sensorineural deafness (OMIM 149200) .It is mapped to, Gene map locus: 13q11-q12 .It is a rare condition, with which few families are affected worldwide [4-10].

It is characterized by knuckle pads, leukonychia, palmoplanter keratoderma (PPK) and sensorineural deafness. However, this syndrome has a considerable phenotypic variability. The clinical features of this syndrome partially overlap with Vohwinkel syndrome and Keratitis-ichthyosis-deafness syndrome [5].

Bart and Pumphrey reported this autosomal dominant condition in a 6-generation family[3].

They disputed whether this complex phenotype could be a monogenic defect with pleiotropic expression [3].

A family reported by Crosby and Vidurrizaga [9] established that keratosis palmoplantaris, probably developing only in older affected persons, is part of the syndrome.

In a multigeneration Polish family with Bart - Pumphrey syndrome, Richard et al [5], reported a novel nonconservative missense GJB2 mutation, segregating with the disorder.

Schwann initially described this condition in families from Poland. Subsequently, cases were also reported from other parts of the world [4-10]. Similar to the condition described by Schwann ; a kindred in which many members had knuckle pads, leukonychia, and deafness due to a lesion of the cochlea, was reported by Bart and Pumphrey [3]. Keratosis palmaris et plantaris was present in some. Male-to-male transmission was thought to have occurred in 2 instances. The presence of leukonychia and the absence of digital constrictions appear to distinguish this disorder from the one listed as 'deafness, congenital, with keratopachydermia and constrictions of fingers and toes' (i.e., Vohwinkel syndrome).

The syndrome is best known currently as Bart - Pumphrey syndrome [4-10].

Although the publication of Jadwiga Schwann on this syndrome [1,2], preceded the publication of Bart and Pumphrey [3] by four years.

Jadwiga Schwann, born in Poland, was precocious. She was working in Szczecin, which is the capital city of the

West Pomeranian Voivodeship in Poland. It is the country's seventh-largest city and the largest seaport in Poland on the Baltic Sea.

The first dermatology researches done in Szczecin, were carried out by Schwann.

Schwann published several papers in dermatology in German and in Polish [11-22]. He has written on different topics in dermatology including mycology and occupational skin diseases [11-22].

I believe that the misnomer Bart - Pumphrey syndrome should be corrected and the syndrome knuckle pads, leukonychia, and sensorineural deafness, should be credited to the right person who reported it first, and should be referred to it as Schwann syndrome.

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JADWIGA SCHWANN AND HER SYNDROME

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Schwann syndrome, also known as Bart-Pumphery syndrome, is an autosomal dominant genodermatosis characterized clinically by knuckle pads, leukonychia and sensorineural deafness and genetically by mutation in *GJB2* gene encoding a gap junction protein, connexin 26. Because Jadwiga Schwann reported this condition in 1963 in German and Polish languages 4 years earlier than the article reported by Bart and Pumphery in N Engl J Med, this disease should preferentially be called as Schwann syndrome. Jadwiga Schwann was a dermatologist in West Poland, who published many articles for both research and clinical topics in the early stage of Polish dermatology.

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