Linear and whorled nevoid hypermelanosis with hyper IgE syndrome

Podłużna i wirowata znamieniopodobna hipermelanoza z współwystępowaniem zespołu hiper-IgE

Joanna Sieniawska[1], Aleksandra Lesiak[1], Wioletta Pietruszewska[2], Anna Woźniacka[1], Joanna Narbutt[1]

[1]Department of Dermatology and Venereology, Medical University of Lodz, Lodz, Poland
[2]Department of Otolaryngology, Medical University of Lodz, Lodz, Poland

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ADDRESS FOR CORRESPONDENCE:
Aleksandra Lesiak MD, PhD
Department of Dermatology and Venereology
Medical University of Lodz
5 Krzemieniecka St
94-017 Lodz, Poland
Phone: +48 42 686 79 81
E-mail: lesiak_ola@interia.pl

ABSTRACT

Introduction. Linear and whorled nevoid hypermelanosis (LWNH) is a disorder of pigmentation characterized by macular hyperpigmentation following the Blaschko lines. Only 50 cases of LWNH have been described so far. Linear and whorled nevoid hypermelanosis may be associated with congenital defects. Underlying chromosomal mosaicism has been demonstrated in only a few published cases. Diagnosis is based on characteristic clinical and histopathological findings.

Objective. Presentation of the first Polish case of LWNH.

Case report. We present a 29-year old Caucasian woman with streaks of reticulate hyperpigmented macules arranged in a whorled pattern over the trunk and extremities. Histopathology showed hyperpigmentation of the epidermal basal layer and prominent melanocytes, without pigment incontinence. Based on clinical and histopathological findings the diagnosis of LWNH was established. Additionally hyper IgE syndrome has been diagnosed in the patient.

Conclusions. In LWNH the therapeutic challenge is the diagnosis and treatment of comorbidities; otherwise it may cause a serious threat to the patient's health.

STRESZCZENIE

Wprowadzenie. Podłużna i wirowata znamieniopodobna hipermelanoza (ang. *linear and whorled nevoid hypermelanosis with hyper IgE syndrome* – LWNH) jest zaburzeniem pigmentacji skóry charakteryzującym się występowaniem przebarwień ułożonych wzdłuż linii Blaschko. Do tej pory opisano ok. 50 przypadków tej choroby. Schorzenie może współistnieć z licznymi wadami wrodzonymi. W kilku opisanych przypadkach stwierdzono występowanie mozaikowatości chromosomalnej. Rozpoznanie ustala się na podstawie charakterystycznego przebiegu klinicznego i badania histopatologicznego.

Cel pracy. Przedstawienie pierwszego w Polsce przypadku LWNH.

Opis przypadku. Prezentujemy przypadek 29-letniej kobiety z ogniskami hiperpigmentacji o układzie linijnym i wirowym zlokalizowanymi na skórze tułowia i kończyn. W badaniu histopatologicznym wycinka skóry stwierdzono hiperpigmentację w podstawnej warstwie naskórka oraz okołonaczyniowe skupienia komórek limfoidalnych z obecnością melanofagów. Na podstawie badania histopatologicznego i obrazu kli-

nicznego ustalono rozpoznanie LWNH. Ponadto u pacjentki potwierdzono współwystępowanie zespołu hiper-IgE.

Wnioski. Istotnym problemem w przypadkach LWNH jest diagnostyka i leczenie chorób towarzyszących, które z nią współistnieją i mogą stanowić poważne zagrożenie dla zdrowia pacjenta.

INTRODUCTION

Linear and whorled nevoid hypermelanosis (LWNH) is a pigmentation disorder characterized by macular hyperpigmentation following the lines of Blaschko. It was first reported by Kalter *et al.* in 1988 [1]. Only 50 cases of LWNH have been described so far, particularly in individuals with high skin phototype. The first skin lesions appear in infancy, usually in the first weeks of life, and gradually worsen. Pigmentations are not preceded by any other skin changes. Stabilization of skin lesions usually occurs after 2–3 years [1–4]. Histopathologically it shows only epidermal melanosis [1, 5–7]. Linear and whorled nevoid hypermelanosis may be associated with congenital defects relating to the nervous, cardiovascular and skeletal systems and ocular anomalies [8–12]. The coexistence of hemiatrophy, Axenfeld-Rieger syndrome, inflammatory linear verrucous epidermal nevus (ILVEN), ichthyosis vulgaris and cerebrovascular malformations is also described [13–16].

Underlying chromosomal mosaicism has been demonstrated in only a few published cases (mosaic trisomy 7, 18 or 20) [17–19]. Hong *et al.* [20] described inversion of chromosome 9 corresponding with LWNH. Furthermore there are also described cases of the occurrence of LWNH inherited in subsequent generations [5–21]. The differential diagnosis includes incontinentia pigmenti, epidermal nevi, hypomelanosis of Ito, Goltz syndrome and Moulin syndrome [22–26]. Diagnosis is based on characteristic clinical and histopathological examination. Dermoscopy shows linear or circular streak-like pigmentations arranged in a parallel manner [27]. However, the rare occurrence of the entity causes diagnostic problems.

OBJECTIVE

We present a case of LWNH, which, according to our knowledge, is the first case reported in Polish literature.

CASE REPORT

We present a case of a 29-year-old Caucasian woman with second skin phototype with progressively increasing streaks of reticulate hyperpigmented macules. The lesions were arranged in a whorled pattern over the trunk and extremities and appeared in the first years of life (Fig. 1 A, B). There was no history of any preceding bullous or verrucous eruption. She was treated in the otolaryngology department due to frequent bacterial and fungal infection of the upper respiratory tract. Moreover, dental defects and aphthae in the oral cavity were observed (Fig. 2). Dermoscopy examination showed linear streak-like pigmentations arranged in a parallel manner (Fig. 3). Histopathological examination of skin lesions revealed hyperpigmentation of the epidermal basal layer and prominent melanocytes, without pigment incontinence. Based on clinical and histopathological findings the diagnosis of LWNH was established. In addition, laboratory tests showed a high level of IgE (2882 kU/l) and defects of innate and acquired immunity. The diagnosis of hyper IgE syndrome was confirmed.

DISCUSSION

We present this case because LWNH is a very rare entity especially among fair skinned subjects. So far only a few cases of the disease have been published. In reported cases association of LWNH with many congenital defects was observed, and in our case LWNH coexisted with immunological defects and hyper IgE syndrome [28]. In the differential diagnosis incontinentia pigmenti, epidermal nevi, hypomelanosis of Ito, Goltz syndrome and Moulin syndrome have to be considered [24–26, 29]. In hypomelanosis of Ito the skin symptoms are associated with malformations of the skeletal system, central nervous system, teeth and hair. A significant percentage of patients have epilepsy, and often mental disorders are found [23]. The skin lesions in incontinentia pigmenti appear after birth, are diverse, and the disease is characterized by successive phases. Initially there appear erythema and blisters, which are converted to verrucous lesions. The next phase is characterized by formation of foci of hyperpigmentations and hypopigmentations. The disease is linked to chromosome X, usually lethal to the male [22]. In

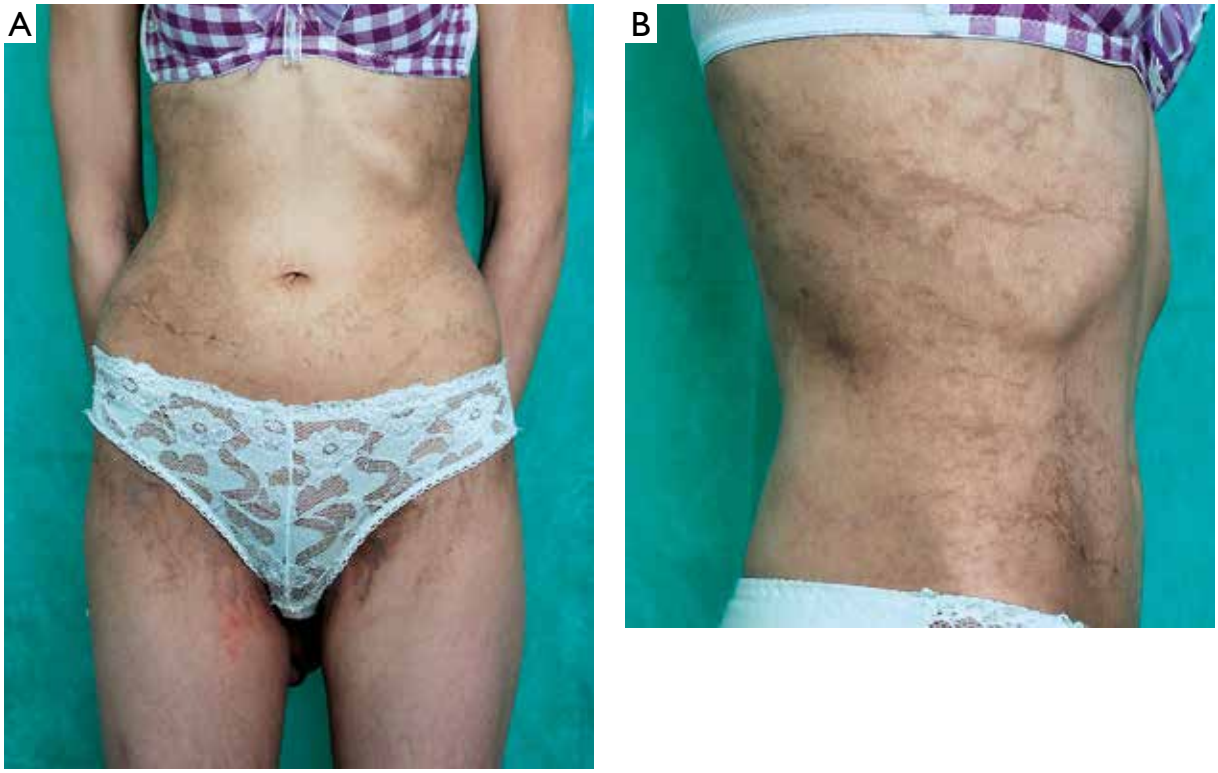

Figure 1 A, B. Skin lesions arranged in a whorled pattern over the trunk and extremities

Rycina 1 A, B. *Zmiany skórne ułożone wirowato na tułowiu i kończynach*

the case of epidermal nevus skin lesions are usually congenital. Initially there appear outbreaks of hyperpigmentation, which later take the verrucous form. Most lesions are linear and do not exceed the central line of the body [24]. In Goltz syndrome, pigmentations are accompanied by other skin changes such as: verrucous papillomas, alopecia, abnormal nail structure, hyperhidrosis, and hyperkeratosis of hands and feet. Comorbidities involved are serious [26].

Moulin syndrome is characterized by the presence of soft linear atrophic, hyperpigmented lesions. Skin changes are usually one-sided, and eruptions occupy the trunk [25]. In our patient, clinical, dermoscopic and histopathological examination showed characteristic features for LWNH, in line with other reported cases [1–16, 27]. Treatment of LWNH does not give satisfactory results. Therapeutic options are limited. The therapy uses chemical peels and 2% hydroquinone [30]. One case was treated with a medium-depth chemical peel regimen using 70% glycolic acid and 35% trichloroacetic acid with no benefit [7]. Our patient did not give informed consent for any topical treatment of her skin lesions.

CONCLUSIONS

In the literature there has not been described coexistence of LWNH with hyper IgE syndrome.

Figure 2. Dental defects

Rycina 2. *Defekty zębów*

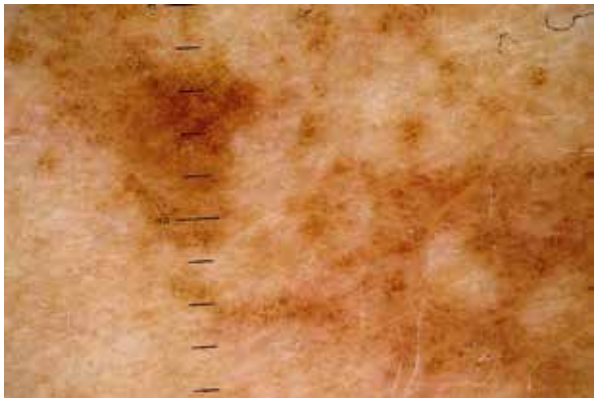

Figure 3. Dermoscopy examination – linear streak-like pigmentations arranged in a parallel manner

Rycina 3. *Badanie dermoskopowe. Linijne, pasmowate przebarwienia ułożone równolegle*

Since LWNH is often associated with the presence of genetic defects and malformations, the most challenging is the diagnosis and treatment of comorbidities; otherwise it may cause a serious threat to the patient's health. According to our knowledge, the presented case is the first case of LWNH in Poland.

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