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GENETIC SKIN DISEASES IN BEAUTY CARE PRACTICE

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Summary

Cutaneous benign tumors and disorders of pigmentation (hipopigmented as well as hyperpigmented spots) are most common skin symptoms in hereditary skin diseases. At the same time these are the basic cutaneous problems which cosmetologists have to deal with in their practice. They are mostly seen at birth or develop during the first years of life. In the majority of cases they indicate the presence or, more often, the increased risk of various systemic disorders development. The fact that such skin lesions are seen several years before systemic alterations occur determine the importance of correct interpretation of such skin symptoms for the complications preventive purposes.

Riassunto

I tumori cutanei benigni e le discromie (aree ipopigmentate ed iperpigmentate) sono sintomi cutanei molto comuni nelle malattie ereditarie della pelle. Allo stesso tempo esse rappresentano i primi problemi cutanei che i cosmetologi devono affrontare nella pratica.

Le discromie si riscontrano maggiormente alla nascita o si sviluppano durante i primi anni di vita. Nella maggioranza dei casi indicano la presenza o, più spesso, il rischio crescente dello sviluppo di varie malattie sistemiche.

Il fatto che tali lesioni cutanee si ritrovano alcuni anni prima che le alterazioni sistemiche si sviluppino, determina l'importanza di una corretta diagnosi di tali sintomi cutanei al fine di prevenire future complicazioni.

Hereditary skin diseases represent the most serious among cutaneous disorders. As a rule genodermatoses are associated with various systemic disturbances. In most cases cutaneous lesions develop a long time before systemic alterations become evident. The importance of correct interpretation of such skin symptoms is beyond doubt. It is a prereqiusite for the early diagnosis which in turn determines further prognosis.

The data discussed below were obtained during the long-term clinical observation of patients with various hereditary skin pathology. All of them developed serious systemic complications, partly as a result of misinterpretation of thier cutaneous lesions prior to their referral to our department.

For the purposes of this article we have subdivided the most common skin changes in two groups: disorders of pigmentation and benign cutaneous tumors.

Analysis of over 150 case histories revealed that such lesions often do not attract the attention of paediatritians, general physicians and sometimes even that of dermatologists. Moreover, the lesions are often being treated as a simple cosmetic problem. Absence of appropriate treatment results in development of systemic complications, inreversible in most cases, including malignant transformation of benignant tumors.

Hyperpigmented spots (cafe-au-lait macules), as well as freckling in auxillary and/or inguinal folds are the earliest skin symptoms of von Reklinghausen disease (neurofibromatosis type I) (5). It is known that various skeletal abnormalities, not equally severe in all cases, are commonly seen in neurofibromatosis. These abnormalities tend to progress with advancing age and sometimes lead to chronic cardiac and pulmonary insufficiency. Fractures of the bones easily develop. In the boy shown on Fig. 1 a very slight trauma resulted in the vertebral column fracture and in a severe 4th degree kyphoscoliosis with subsequent cardiovascular and respiratory impairment and pronounced physical retardation.

Multiple lentigines, predominantly localized periorificially, on mucous membranes, digits,

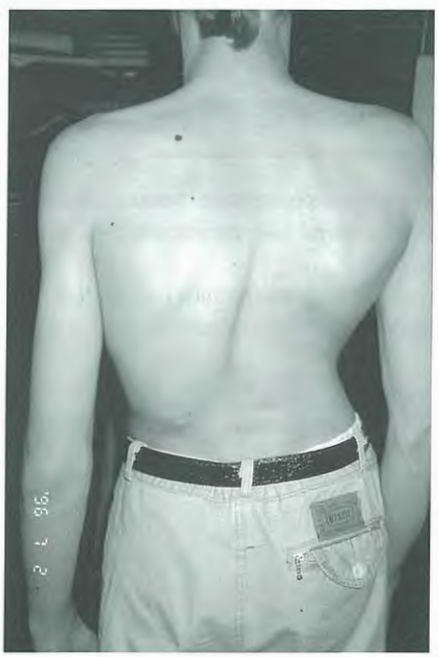

Fig. 1

Fig. 2

palms and soles are also seen at birth or appear during the first years of life. They are asymptomatic. In some patient these spots were diagnosed as moles which did not require further medical observation. But being a symptom of Peutz-Jegers-Touraine syndrome (periorificial lentiginosis) (4), they are the markers of the small and large intestine polyposis which develops several years later with the increased risk of malignant transformation of polyps into adenocarcinoma.

Maple leaf - like hypopigmented spots, seen in children, are the earliest cutaneous signs of Burneville-Pringle disease (Fig. 2). As we could see, some of the patients consulted at our department due to the previous misdiagnosis had an advanced epilepsy and pronounced mental retardation. It should also be remembered about the increased incidence of malignant brain tumors development as well as cardiac and renal complications in these patients. The vital importance of therapeutic investigation of such children before the systemic pathology develops is obvious.

Cutaneous changes in the girl, shown on Fig. 3 were diagnosed as multiple pigmented naevi. But lentigines of generalized pattern are the earliest symptom of LEOPARD-syndrome. Patients must be under clinical supervision to prevent the development of cardiovascular, neurologic complications etc.

Pigmented spots in neurofibromatosis I type are seen at birth and in some patients almost 15-20 years before the development of neurofibromas, which sometimes, especially in case of plexiform neurofibromas may transfer into neurofibrosarcoma. The misdiagnosis in patient shown of Fig. 5 and removal of just a single dermal neurofibroma at a beauty care center resulted in development of thousands of neurofibromas which covered most of body surfaces including the face.

Inherited cutaneous tumors, especially if they develop in early childhood may be helpful in preventing serious systemic complications. For instance, it is known that in overwhelming majority of patients with neurofibromatosis type 2 bilateral acoustic neuromas result in deafness on average at the age 20-30 (6). That is why it is very important for the patient to be under medical supervision, in case cutaneous schwannomas are diagnosed in association with multiple cafe-au-lait spots.

Spitz nevus (Fig.4) is not a genodematosis per se, but we have seen several cases of congenital spindle and epithelioid cell nevi, that were treated with electrocoagulation as angiomas when solitary, and as molluscum contagiosum when multiple in infants. As it is, Spitz nevus is one of the most common simulators of malignant melanoma of the skin, both clinically and histologically (1,3). However, when it recurs after such unnecessary medical manipulations (with a great deal of inflammation, as we have seen), it may acquire more atypical features that become misleading for the pathologist.

We have suggested that multiple cutaneous leiomiomas (Fig. 5) indicate the increased risk of uterine myoma. Such association was present in all family cases observed by us. It is very impor-

Fig. 3

tant to say that all out patients underwent hysterectomy at child-bearing age. As cutaneous changes are seen several years before the uterine myoma develops, the importance of timely diagnosis cannot be overestimated.

The presence of multiple sebaceous tumors (adenomas as a rule) should rise suspicion and prompt examination should be made to reveal the internal malignancies (gastrointestinal most commonly), which usually precede cutaneous lesions.

Thus certain skin lesions can be helpful in correct diagnosis of internal pathology, which, if made in time, improves further prognosis and course of the disease.

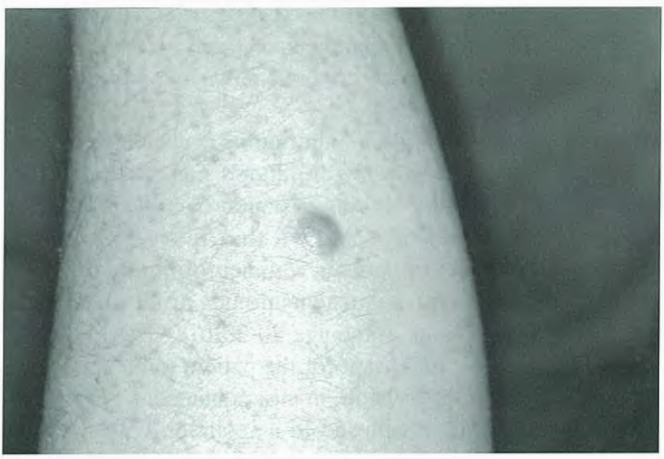

Fig. 4

Fig. 5

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