The CHILD-Syndrome—Congenital Hemidysplasia with Ichthyosiform Erythroderma and Limb Defects. A Case Report

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A presentation of a 3-year-old girl with a congenital hemidysplasia of the left side together with ichtyosiform erythroderma, the so-called CHILD-syndrome. A short review of the syndrome is given. *Key words: X-linked dominant gene-defect, Lyon-effect.* (Received September 9, 1983.)

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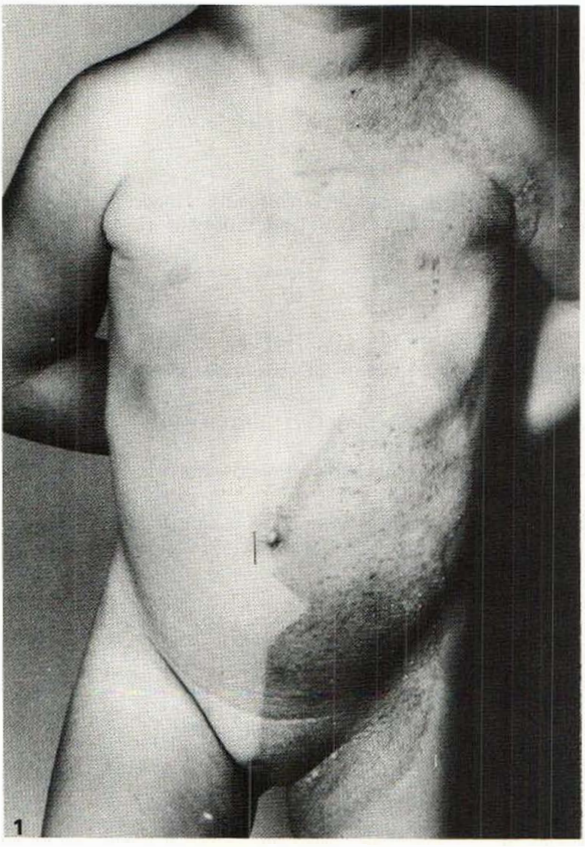
In 1980 Happle et al. (1) proposed the term "CHILD-syndrome" for what was previously named "congenital unilateral ichthyosiform erythroderma". The reason for introducing the new name was to give a better description of the syndrome and to focus on new genetic aspects.

In 1982 Happle (2) had collected a total number of 25 published cases—all but one of female sex. The present paper describes another case of the CHILD-syndrome, probably a mutant.

CASE REPORT

A 3-year-old girl was admitted for the first time to our clinic at the age of three months. The family history was negative and her parents not related. A 6-year-old sister was without symptoms. Her birth



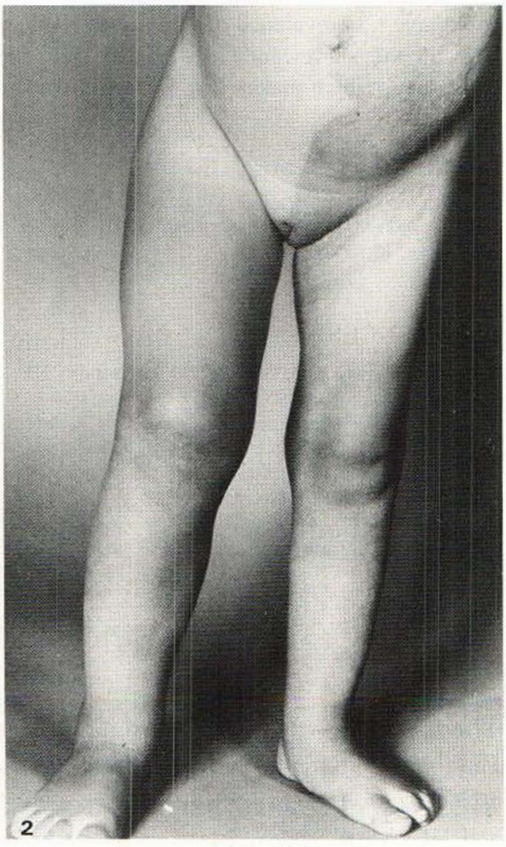


Fig. 1. The skin eruptions are localized only to the left side of the body and extremities.

Fig. 2. The shortness of the left leg is clearly seen.

weight was 2790 g and her length 48 cm. The mother had no history of abortions, and the present pregnancy and birth was without complications. At birth atrophy of the left-sided extremities and syndactyly of the fourth and the fifth finger was observed. When examined by us we found the skin of the left side of the body and the left-sided extremities hyperkeratotic and erythrodermic and diagnosed the case as a "congenital unilateral ichthyosiform erythroderma" or CHILD-syndrome (Fig. 1). The skin was primarily treated with neutral ointments containing salicylic acid 1%.

A certain improvement was observed during the first few years. However, at the age of three exacerbation took place, which motivated introduction of systemic treatment with Tigason® (Etretinate). All skin symptoms were still strictly located to the left side of the body and the left leg was found to be 6 cm shorter than the right one (Fig. 2). At present it is still too early to estimate the results of this treatment.

Histopathology

In a punch-biopsy from the left axillary region a thickened epidermis with acanthosis and a lamellar hyperkeratosis with prominent parakeratotic foci was found. The finding was most pronounced over the elongated papillae. The keratinisation superficial to the rete ridges was orthokeratotic, and there was a thickened granular layer. No ballooning was observed. The acanthotic epidermis contained several dyskeratotic cells, and in the basal layer there was spongiosis. The papillary dermis revealed slight oedema and diffuse infiltration of lymphocytes. The histological changes were similar to those previously described for recessive, congenital ichthyosiform erythroderma.

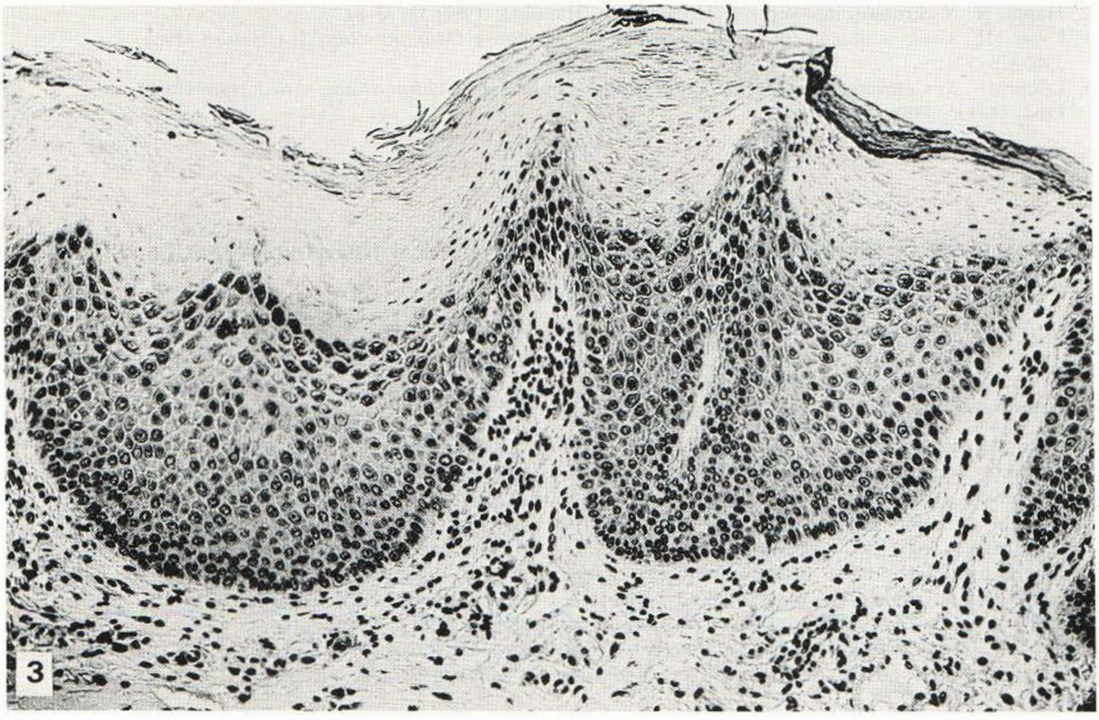


Fig. 3. Punch-biopsy from the left axillary region. The papillary pattern resembles psoriasis and so the foci of parakeratosis, but not the prominent granular layer. In the papillary dermis small infiltrates of lymphocytes perivascular. (Van Gieson stain, ×125.)

DISCUSSION

The inheritance of this abnormality has previously been described as an autosomal recessive transmission. Happle et al. (1) proposed an X-linked dominant gene-defect lethal in the hemizygote male foetus.

Furthermore, in 1982 Happle (2) proposed an explanation for the unilateral distribution of the defects, referring to the so-called "Lyon-effect" (3, 4). The hypothesis is, that during the first weeks of foetal life an inactivation of one of the X-chromosomes in all somatic female cells will occur. This may happen at different stages. If it occurs at an early stage, half of the body cells will receive the normal X-chromosome from the father, and the other half the defect chromosome from the mother. Thus, in one half of the body the cells will have the normal gene from the father and in the other half the defect gene from the mother.

The case of the CHILD-syndrome published here is most likely to be a mutant. In her family there are no other cases or abnormalities.

Her skin problems are obviously grave, but the most serious abnormality seems to be the retarded growth of her left-sided extremities.

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