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PSEUDO-PSEUDO-HYPOPARATHYROIDISM AND BASAL-CELL NEVUS SYNDROME

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SUMMARY

A case is described of pseudo-pseudo-hypoparathyroidism, with the following symptoms: round face, large forehead, relative backwardness, short stature, relative obesity, congenital muscular torticollis, cervical fused vertebrae, spina bifida occulta cervicalis, bifid rib, bilateral brachymetacarpy IV. Some of these also occur in the basal-cell nevus syndrome. The patient does not however show a basal-cell carcinoma, nor mandibula cysts.

The following case study is that of a pseudo-pseudo-hypoparathyroid woman. The subject also shows, however, some anomalies which can be seen in the basal-cell nevus syndrome. This case illustrates at the same time the possible nosological relation between those two rare genetic syndromes.

HISTORY OF THE ILLNESS

D.A., 35, comes to the examination because of chronical backaches.

The clinical examination does not give any precisions. Normal skin, black hair, light backwardness. The patient has been married for seven years with a healthy man and has no children. Normal menses after a normal menarche. She broke her right humerus in 1961 for which various interventions with osteosynthesis were necessary. Subject has a striking habitus: short in stature (151 cm); relative obesity (73 kg). She has a slight wryneck to the left: this, after an operation in 1944 for a congenital muscular torticollis. Her face is round. The neck is strikingly short, with low implanted hair. The hands are short, with an abnormally short 4th finger, bilaterally (Fig. 1).

Urine, peripheral blood, uremia, cholesterolemia, and alkaline phosphatases are normal. Calcemia 8.8 and 8.7 mg%; phosphatemia 1.6 and 2.3 mg%; PBI 8.7 γ%; basal metabolism normal. The glucose-tolerance test is abnormal, with a glycemia peak till 220 mg%.

RADIOLOGICAL EXAMINATIONS

Skull: light skull asymmetry.

Cervical column: congenital fused vertebrae $C1$-$C3$: spina bifida of $C6$-$C7$.

Thoracic column: spina bifida from $D1$ till $D3$; abnormal form of the corpora $D4$ and $D5$.

Forearms: light radius curvus, bilaterally no exostoses.

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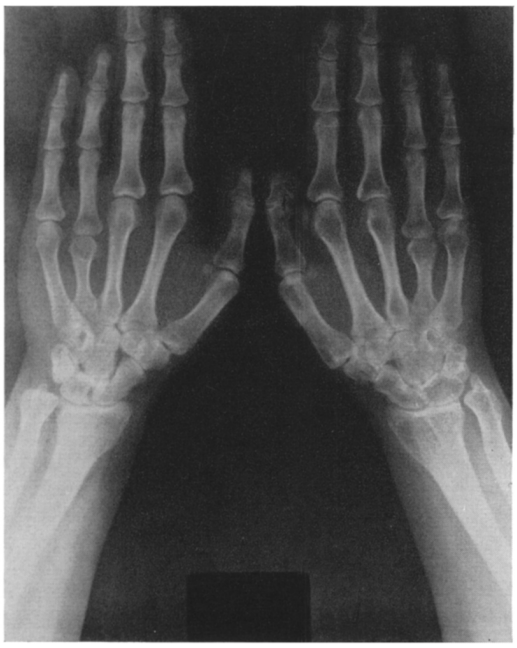

FIG. 1. *Bilateral Brachymetacarpy IV.*

Hands: brachymetacarpy IV, bilaterally (Fig. 1). Normal phalanges.
Lumbar column: normal, no spina bifida, lumbarisation of *S*1.
Thorax: bifid rib on the left.
Abdomen: normal, no lithiasis visible.
Pelvis: normal, no exostoses.
Feet: normal, no brachymetatarsy.
IV. pyelography: bilateral normal secretion and excretion. Normal arborisation.

EXAMINATION OF THE FAMILY

The mother of the patient had normal hands, although bilateral shorter 4th toes. Normal stature. She died one year before our study.

The father died from a stomach carcinoma. He had a short stature, tough had normal hands and feet.

No data could be found about grandparents, uncles, and aunts.

The patient's elder sister is tall, with normal hands and feet. She has been married for fifteen years and has no children.

The second sister is under treatment for sarcoidosis of the lung. Normal stature. Positive metacarpal sign to the right, due to a slight brachymetacarpy IV. Normal feet and skull. She has two normal children. No known consanguinity in the family.

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DISCUSSION AND CONCLUSIONS

The small stature, relative obesity, round face, brachymetacarpy IV, moderate backwardness, brachymetatarsy of the mother, are typical for the diagnosis of pseudo-pseudo-hypoparathyroidism (Albright et al. 1952, Forbes 1962, Goeminne 1963 and 1965, Klotz et al. 1962, Mann et al. 1962, Schwarz 1964).

Ectopic calcifications and calcifications in the basal ganglia are however lacking. An Ellsworth-Howard test has not been done. An earlier tetany or hypocalcemia could not be proved.

This is the fourth description known by us about this syndrome in Belgium, after the previous observations of Nagant de Deuxchaisnes et al. (1960), Goeminne (1965), and De Schrijver and Van Nevel (1968).

The round face, backwardness, brachymetacarpy IV, bifid rib, cervical fused vertebrae, and spina bifida occulta cervicalis, remember, on the other hand, the basal-cell nevus syndrome (Binkley and Johnson 1951, Gorlin et al. 1965, Berlin et al. 1966), a rare affection, about a hundred cases of which have been described from 1951 to 1957, inherited as a dominant with high penetrance. Some important skin and skeletal symptoms are however lacking, such as basal-cell carcinomas, palmo-plantar dyskeratosis, and especially mandibular cysts. Calcifications of the falx cerebri are also lacking. The skin symptoms of this syndrome, however, usually occur after the skeletal symptoms; most of the time they are present before age 40.

The basal-cell nevus syndrome mainly consists of skin and skeletal symptoms, besides neurological, sexual, and ophtalmologic anomalies. The skin symptons are especially the multiple basal-cell carcinomas of the face and the palm of the hand and the footsoles. Epithelial cyst also appear. The bone lesions especially consist of: bone-cysts in the mandibula, a small sella turcica, synostosis of the ribs, bifid ribs, cervical ribs, spina bifida occulta of the cervical and high thoracic region, fused vertebrae, cyphoscoliosis, and brachymetacarpy IV and V. Teath disturbances with an early caries also frequently appear. Neurological symptons can consist of a backwardness, EEG disturbances, ectopic calcifications in the falx cerebri, agenesis of the corpus callosum, and sometimes medulloblastomas. The great majority of the patients have a round face with a large forehead. Sexual anomalies sometimes consist of hypogonadism, whether or not with cryptorchidism in men, and in women always with solid benign ovarian fibromas that do not seem to influence fertility. Eye symptoms are especially hypertelorism, dystopia canthorum and sometimes internal strabismus.

In some of the patients with a basal-cell nevus syndrome, an Ellsworth-Howard test was performed resulting positive, which means that a parathyroid infusion cannot suppress parathyroid function, i.e., there is no increased phosphaturia (Block and Clendenning 1963, Gorlin et al. 1965). Patients with pseudo-hypoparathyroidism show the same phenomenon.

Short size and brachymetacarpy IV also occur in the metacarpal brachydactyly type Bell, E_2, in which other anomalies, except for short stature, are usually lacking. A short size and brachymetacarpy IV are also classic symptoms in gonadal dysgenesis.

The following conclusions may be drawn:

1. One has to investigate carefully the hands of a patient with a short stature, relative obesity, and backwardness.

2. A complete endocrine and radiologic skeleletal examination is necessary in short sized patients who have relative obesity, backwardness, and brachymetacarpy.

3. This case also illustrates the possible nosological and genetic relation between psudo-pseudo-hypoparathyroidism and basal-cell nevus syndrome.

4. A positive metacarpal sign of Archibald may be a *forme fruste* of pseudo-pseudo-hypoparathyroidism.

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RIASSUNTO

È stato descritto un caso di pseudo-pseudo-ipoparatiroidismo che presentava, fra l'altro, fusione delle vertebre cervicali, spina bifida occulta cervicalis, costa bifida — anomalie che si riscontrano anche nella " basal cell nevus syndrome ".

Résumé

Nous avons décrit un cas de pseudo-pseudo-hypoparathyréoïdie, présentant les anomalies suivantes: des vertèbres cervicales fusionnées, une spina bifida occulta cervicalis et une côte en fourche. Ces anomalies se rencontrent aussi dans le " basal cell nevus syndrome ".

Zusammenfassung

Beschreibung eines Falls von Pseudo-Pseudo-Hypoparathyreoidismus, der u.a. eine Verschmelzung der Halswirbel, eine spina bifida cervicalis occulta sowie eine costa bifida aufwies. Diese Anomalien finden sich auch beim " basal cell nevus syndrome ".

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