Craniometaphysial Dysplasia with Leukoencephalopathy

A Case Report

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Summary. A case of craniometaphysial dysplasia with extensive degeneration of the cerebral white matter is presented. The cortex of the swollen part of the long bones was quite thin and there was striking trabecular atrophy. Both the vault and base of the skull showed marked thickening and sclerosis, leaving no interlaminal zone.

It is probable that the diffuse degenerative change of the cerebral white matter with gliosis bears some resemblance to that produced by a circulatory disturbance of the great vein of Galen. Due to the narrowed foramen magnum, deformed atlas and axis, and the surrounding postoperative scar, the upper cervical cord was compressed, markedly atrophic and degenerated. Other segments of the cervical and thoracic cord displayed secondary wallerian degeneration and focal neurolytic lesions in the white matter. At the level of Th11 there was a pencil-like malacic lesion, suggesting an apparent interference of circulatory disturbance due possibly to the deformed vertebral column.

Key words: Pyle's disease – Craniometaphysial dysplasia – Leukoencephalopathy – Spinal cord compression.


Es wird angenommen, daß die diffusen degenerativen Veränderungen der weißen Hirnsubstanz mit Gliose Ähnlichkeiten haben mit jener, wie sie bei Abflußbehinderung in der Vena cerebri magna galeni beobachtet wird. Wegen des engen Foramen occipitale magnum, dem deformierten Atlas und Epistrophæus und den umgebenden postoperativen Narben erscheint das obere Halsmark komprimiert mit ausgeprägter Atrophie und degenerativen Verände-
rungen. In anderen Anteilen des Hals- und Brustmarkes finden sich sekundäre
Waller'sche Degenerationen sowie fokale Erweichungen der weißen Substanz.
Auf Höhe von Th11 findet sich eine Verschmälerung des Markes auf Bleistift-
dicke bei lokalisierter Malazie. Letztere dürfte eine Folge der hier deformierten
Wirbelsäule sein.

Introduction

Pyle's disease was originally described in 1931 as a rare disease causing an
abnormal swelling of the epiphysis of long bones. Association of the sclerosis of
the base of the skull and facial deformity with this disease is called cranio-
metaphysial dysplasia (Jackson, 1954), and its familial occurrence is also
suggested (Backwin and Krida, 1937).

When the disease intensely involves the base of the skull, various symptoms
and signs of cranial nerve impairment such as optic and facial nerve atrophy and
acoustic disturbance are associated, probably due to stenosis of the basal foramen
or disorder of transmission due to otosclerosis.

This disease is not considered to be rare in Europe and the USA but, only
eight cases have been recorded in Japan. Autopsy findings of the brain were
reported only in one case by Millard (1967).

The present paper describes neuropathological features noted in a case of
craniometaphysial dysplasia with marked changes in the brain and spinal cord.

Case Report

A woman, aged 25 years, whose parents were cousins, was noted, two months after a normal
delivery, to lack sensation to light. A diagnosis of optic nerve atrophy of unknown etiology was
made. Her growth, mental development and motor functions were normal until she was about
13 years old when she developed a weakness of the left hand, a disturbance of gait and sudden
worsening of her vision in September 1963. Within a year she unable to stand and had to be
admitted to a public hospital. Her weight was 25 kg and height 131 cm. There was an abundance
of hair over the body, atrophic lesions of the skin and palpable bony overgrowth at the joints of
the four extremities. X-rays revealed flask-like hypertrophy of the metaphysis of all the long
bones, marked osteosclerosis of the skull with obliteration of the paranasal sinuses, hypoplasia
of the atlas and axis, and narrowing of the foramen magnum. Views of the optic canals were not
obtained. The condition advanced rapidly to spastic tetraplegia and a laminectomy of the atlas
and widening of the foramen magnum were performed under the diagnosis of Pyle's disease.

She was admitted to the Toho University Hospital at the age of 21 because of ileus. She was
130 cm tall, blind, aphasic and spastic. At the time of her final admission August 29, 1975 she
was decerebrate and had advanced decubitus and bronchopneumonia. X-rays were similar to
those described above (Figs. 1 and 2). Laboratory examinations disclosed marked hypoproterinem-
aemia, anemia and slight adrenocortical hypofunction. She died on November 18, 1975.

Autopsy Findings

The body was small and markedly emaciated, measuring 125 cm in length and
weighing 25 kg. All organs were atrophic and the direct cause of death was