A CASE OF PHOCOMELIA ASSOCIATED WITH SEVERE MENTAL DEFICIENCY

By

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PHOCOMELIA is a congenital malformation of the osseous system characterized by the absence or extreme smallness of the arms, forearms, thighs and legs, producing a pronounced shortening of limbs. The hands and feet, which may be fairly normal in appearance and development, seem inserted more or less directly into the shoulders or pelvis of the patient. As the micromelic appendages resemble the physical outlines of a seal's flippers, the condition is known as phocomelia.*

Sometimes, however, only the arms or the legs are affected and less uncommonly, a single arm or leg with all the other limbs otherwise normally formed. The hands and feet, however, may show numerous congenital anomalies such as disproportionately short, long, supernumerary and webbed fingers or toes.

This exceedingly rare bone anomaly is related to pre-natal factors arising during the early phase of foetal development which prevent the laying down of certain parts of the osseous system. The cranial vault, the vertebral arches, the pelvic and the shoulder girdles, may likewise manifest serious structural deficiencies.

Short references to phocomelia have been made by Church (1911), Aschoff (1928) and Kaufmann (1929) but the most descriptive record with photographs has been contributed by Patten (1946). None of the above-mentioned authors, however, have investigated psychologically the mental condition and intellectual state of their patients.

A case of phocomelia has recently been admitted to this Mental Deficiency Institution. She is a white baby girl aged 3·7 years chronologically, having a mental age of 8·2 months and an intelligence quotient of 19 being classified as belonging to a low-grade type of mental defect (Idiot).

She is of Caucasian race, and comes from a non-farm, rural environment. She is the last born of two siblings, her older brother being of normal physique, intelligence and in good health.

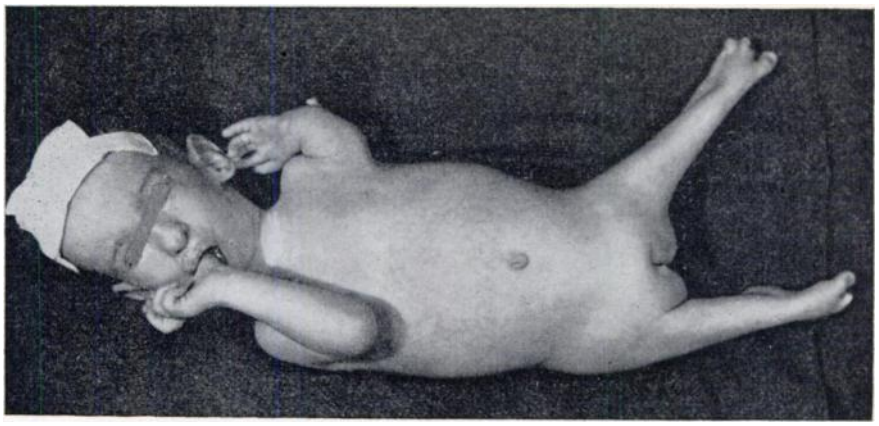
The mother, who is a diabetic with a history of psychosis, was 35 years old and the father, who has a peptic ulcer, 43 at the birth of the patient. Histories of alcoholism in the paternal, and psychosis in the maternal grandparents have also been recorded.

PERSONAL HISTORY

The patient was born at eight months. The mother felt well during the pregnancy, and labour was stated as having been uneventful. The baby weighed 5 pounds at birth and had feet and a left arm growing out of the

* Phocos, Greek for seal; melos, Greek for limb.

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A case of Phocomelia Associated with Idiocy.

lower torso and left upper shoulder-chest region, respectively. She had a hare lip and a cleft palate which were recently repaired surgically. Otherwise she was a healthy baby, breathing without difficulty, and moving about in the crib playfully. No other information is available.

Physical Examination

Skin: Clear.

Scalp: Normal.

Hair: Fair and fine.

Eyes: Blue; palpebral fissures are narrow, with a certain degree of slanting towards the outer angles.

Nose, lips and palate plastically repaired.

Lungs: Clear to A. and P.

Heart: No murmurs heard and cardiac outlines normal.

Neuro-muscular system: She is right-handed with pareses of both lower extremities and left upper limb. The grip of the right hand is normal, reflexes of left biceps active, right triceps sluggish. Left biceps and triceps reflexes are absent. The upper and lower abdominal reflexes, both left and right, are active, and the plantar reflexes are normal bilaterally.

Special Data

Weight on admission: 14 lb. 6 oz. (Since her admission three weeks ago, she has gained over 1 lb. 4 oz. in weight.)

Length: 22 inches.

Cranial circumference: 17½ inches.

Length of normal right arm and forearm: 9 inches, shortened left arm and forearm 5 inches. The lower limbs measure only 7½ inches, including feet.

Torso-Leg Ratio: 19 inches/14½ inches.

The inner aspect of the skin of the shortened arm is adherent along the axillary line to the skin of the outer chest wall; the left elbow joint is movable and the left forearm overly shortened.

Feet: There are syndactylies involving the second and third as well as the fourth and fifth toes of both feet, which are unusually small. A pilo-nidal sinus is seen in the ilio-coccygeal region.

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Roentgenological Report

There are most amazing osseous defects in this child. The left humerus, radius and ulna are underdeveloped and deformed but the bony outlines of the right upper extremity are normal. Both femurs are hypoplastic and of abnormal shape and both tibiae and fibulae are absent. The facial bones, particularly the sphenoid, ethmoid, maxillary and mandibular structures, are rather smaller than usual, and there is a cleft in the bony palate. The left shoulder blade is hypoplastic, of irregular outline, showing some defect in its centre. The cranial vault exhibits slight digital markings with some thinning of inner table.

Orthopaedic Consultation

Congenital absence, multiple in extremity bones, left-upper and both lower limbs markedly shortened. Cleft palate, hare lip repaired. Later in childhood, if it appears indicated at that time, prosthetic considerations may be given.

Ophthalmological Consultation

OD Pup. 3^5 reg. and act. Homatropine instilles. Media clear. Discs clear, good colour. Vessels and maculae normal. OS similar to OD. EQM normal. No pathology seen.

Psychological Consultation

Gross motor patterns fall between the eight- and sixteen-week levels. Standing, unable to support any weight, below twelve weeks; supine, pulled to sitting, complete head lag twelve weeks; prone, head zone III 16-week level. Adaptive patterns are somewhat higher, i.e. radial palmar grasp 28 weeks, and transfers adeptly 28 weeks. No verbalization is elicited. Sphincter control is absent, cannot sit up or feed herself. Chronological age: Three years seven months; mental age, eight and two-tenths of a month, intelligence quotient 19. Classification: Idiocy.

Summary and Diagnosis

In view of the above findings, it appears that this child manifests the congenital malformation described as phocomelia which is complicated by a severe degree of intellectual defect (Idiocy), cleft palate, hare lip, skull anomalies, pedal syndactylism and pilo-nidal sinus.

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