

Hypoplasia of Nails and Phalanges : A Teratogenic Manifestation of Phenobarbitone

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Anticonvulsants such as diphenylhydantoin and sodium valproate when given during pregnancy are known to induce certain types of malformations in the fetus(1). As against this, drugs such as phenobarbitone and carbamazepine are considered relatively safe in pregnancy(2).

We are reporting herewith a case of malformations in a child secondary to phenobarbitone consumed by the mother during pregnancy.

Case Report

A 6-year-old girl presented with a history suggestive of repeated lower respiratory tract infections and dyspnea on exertion from two years of age.

The child was born of a non-consanguineous marriage. Other two elder sibs were

normal. There was no family history of any inherited diseases or congenital anomalies, including nail hypoplasia. The mother had an intracranial tuberculoma, diagnosed ten years ago, which required excision and eighteen months of antituberculous therapy. She was advised longterm phenobarbitone therapy in the dose of 90 mg per day. She had continued taking phenobarbitone during the pregnancy and was seizure-free throughout the pregnancy. No other anticonvulsants were taken at any time. There was no history of exposure to other teratogens or intrauterine infections. On thorough examination and investigations, the patient was found to have a large perimembranous ventricular septal defect with moderate pulmonary hypertension which was confirmed on two dimensional echocardiography. She had dysmorphic features like microcephaly (head circumference 43 cm), metopic ridge, hypertelorism, epicanthal folds, broad alveolar ridge, (Fig. 1), absence of distal phalanges of fingers and toes with hypo-plastic nails, (Figs. 2 and 3), pilonidal sinus and bilateral mild congenital talipes equino varus (CTEV). The patient also had psychomotor retardation with an IQ of 58 and a social quotient (SQ) of three and half years. There was no evidence of intrauterine infection on clinical examination.

There were no renal anomalies detected on ultrasonography and skeletal survey was normal, barring the absence of distal phalanges of fingers and toes. The dermatoglyphics revealed a normal pattern. Karyotyping of the patient was normal.

Discussion

Anticonvulsants are an important group of teratogens. Diphenyl-hydantoin is known to cause the well-known "fetal-

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Fig. 1. Patient's photograph showing dysmorphic features.

hydantoin syndrome" with nail hypoplasia an important hallmark(3,4).

Phenobarbitone is very rarely known to cause teratogenic effects such as cleft lip, cleft palate, congenital heart defects (TOF, PDA, VSD)(5,6) and increased risk of intracranial tumors(7). Coagulation defects and hypocalcemia in the neonate are also known(8,9). However, nail hypoplasia with absence of distal phalanges have not been reported.

The present case had prolonged and continuous antenatal exposure to phenobarbitone, and fetal expression in the form of digital and nail hypoplasia. No anticonvulsant drug other than phenobarbitone



Fig. 2. Fingers and toes with absence of distal phalanges and nail hypoplasia.

was used, and there was no family history of digital hypoplasia. In all probability it seems that phenobarbitone caused these congenital anomalies in our patient.

Since there were no other obvious etiological factors which could have caused these congenital defects, we presume that they were due to antenatal use of phenobarbitone.

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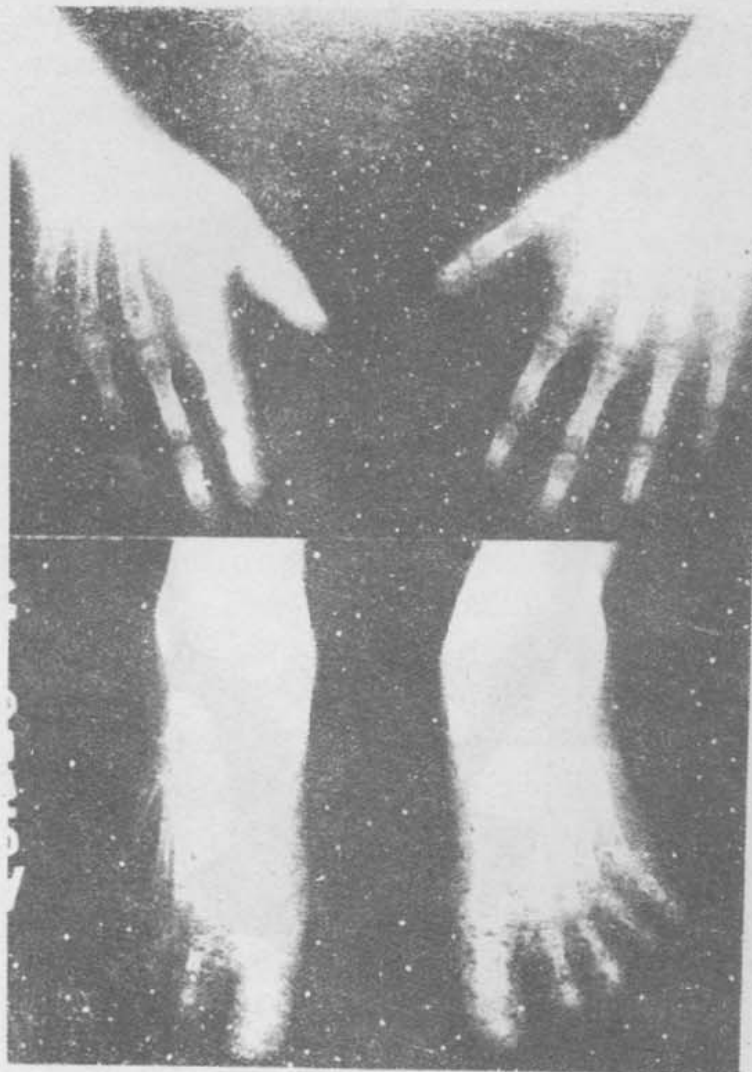


Fig. 3. Roentgenograph showing absence of distal phalanges.

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Lipoid Proteinosis (Urbach-Wiethe Syndrome) with Dwarfism

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Urbach-Wiethe syndrome also known as lipoid proteinosis or hyalinosis cutis et

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