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Case Report

Microcephalic osteodysplastic primordial dwarfism (MOPD) type I with severe anemia and MRI brain findings of MOPD type II

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ABSTRACT

We report a 4 month old male, 4th in order of birth of healthy consanguineous Egyptian parents with typical characteristics of microcephalic osteodysplastic primordial dwarfism most probably belongs to type I (MOPD I). The patient had intrauterine growth retardation, sparse scalp hair, sparse eyebrows and eyelashes, high arched palate, micrognathia, low set ears, short neck, clenched fists, groove between thumb and palm of hand, arachnodactyly, flexion contractures of elbow and knee. He also had thin dry skin with marked decreased subcutaneous fat and prominent superficial veins over chest and abdomen and mild hypertrichosis over lower back and buttocks. However, the patient had severe anemia and MRI brain findings revealed global hypovolemic brain changes in the form of dilated ventricles and widened cortical sulci, multiple old vascular insults and aneurismal dilatation of right internal carotid artery (ICA) which are consistent with MOPD II.

1. Introduction

Primordial dwarfism is a very rare form of dwarfism beginning in early stages of intrauterine life and results in a smaller body size in all stages of life [1]. Primordial dwarfism is a very heterogeneous group of disorders and it has been classified into three main types: Seckel syndrome, microcephalic osteodysplastic primordial dwarfism (MOPD) type I/III and type II [2].

Microcephalic osteodysplastic primordial dwarfism (MOPD) is a syndrome characterized by the presence of intrauterine growth restriction, post-natal growth deficiency, microcephaly and a similar phenotype to Seckel syndrome. This condition was initially described by Majewski et al., [3] who characterized three distinct syndromes which he named microcephalic osteodysplastic primordial dwarfism types I, II and III. Majewski et al. also established the difference between these and Seckel syndrome due to the severity in the growth retardation, the presence of bone abnormalities and mild or absent mental retardation [4–7].

We report a case with the typical features of microcephalic osteodysplastic primordial dwarfism type I who had in addition some unreported features most probably belongs to MOPD II after taking consent of the parents.

1.1. Case report

A 4 month old male, 4th in order of birth of 1st cousin consanguineous marriage. The patient was delivered at full term by cesarean delivery after uncomplicated pregnancy with no history of fever, drug intake or smoking by the mother. His birth weight was 1.6 kg (<5th percentile). The patient was referred to the Genetics Clinic, Pediatric Hospital, Ain Shams University complaining of poor weight gain. At birth the patient was admitted to neonatal intensive-care unit (NICU) for 23 days for jaundice. At the age of 1.5 month he developed vomiting with every breast feeding not associated with fever or diarrhea. The mother added artificial milk formula without improvement. At the age of 75 days, he was readmitted to NICU for dehydration and blood transfusion was given once for anemia. At the age of 3 months, he was readmitted to hospital for refusal of oral intake and vomiting and a nasogastric tube was applied for feeding. He developed anemia and blood transfusion was given once more during admission. He also had attacks of convulsions and started Tiratam therapy. Gradually there was improvement of oral intake and the patient was discharged from hospital and he could gain 400 g within one month.

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Family history revealed previous sib death at the age of 40 days with the same condition who also developed anemia. He had two other healthy sibs. Both parents were normal.

On examination, his weight was 2.600 kg (below 5th percentile), his length was 41 cm, (below 5th percentile), his span was 41 cm, and his skull circumference was 31.5 cm (below 5th percentile). The patient had sparse hair on the posterior part of the scalp and absent hair on anterior part of the scalp, sparse eyebrows and eyelashes, high arched palate, micrognathia, low set posteriorly rotated ears, short neck, clenched fists with a groove between thumb and palm of hand, arachnodactyly, flexion contractures of elbow and knee joints, thin dry skin with marked decreased subcutaneous fat and prominent superficial veins over chest and abdomen (Figs. 1, 2). He also had mild hypertrichosis over lower back and buttocks and thin wrinkled skin (Fig. 3).

Abdominal examination revealed bilateral inguinal hernia (Fig. 4). Cardiac, genital and neurologic examinations were normal.

Abdomino-pelvic ultrasonography and ECHO cardiography were normal. Extended metabolic screen, Karyotype and barium swallow were also normal.

Complete blood picture revealed HGB 10 g/dL, platelets 478 000/cm, serum iron 87 μg/dl (normal range 50–120), serum total iron binding capacity 266 (normal range 250–450), serum transferrin 32 (normal range 20–50). Kidney and liver function tests were normal.

Plain X-ray of the chest revealed bell-shaped thoracic cage with eleven pairs of ribs. Lateral view of the spine revealed mild lumbar kyphosis. The lumbar vertebrae are oval in shape with small central peaking. Plain X-ray of the left wrist revealed absence of carpal ossific centres (Fig. 5).

MRI brain revealed aneurysmal dilatation of the internal carotid artery associated with multiple old infarcts. Ventricular dilatation is also seen indicating atrophic brain changes (Fig. 6).

2. Discussion

Microcephalic osteodysplastic primordial dwarfism (MOPD) has three subtypes I, II, III. Although MOPD I and III were originally described as two separate entities on the basis of radiological criteria mainly in long bones and pelvic bones, later reports confirmed that the two forms represent different expression of the same syndrome. MOPD I commonly has hair thinness in scalp, eyelashes and eyebrows, protruding eyes, prominent nose with a flat nasal bridge, small low-set ears, micrognathia, small chin and short

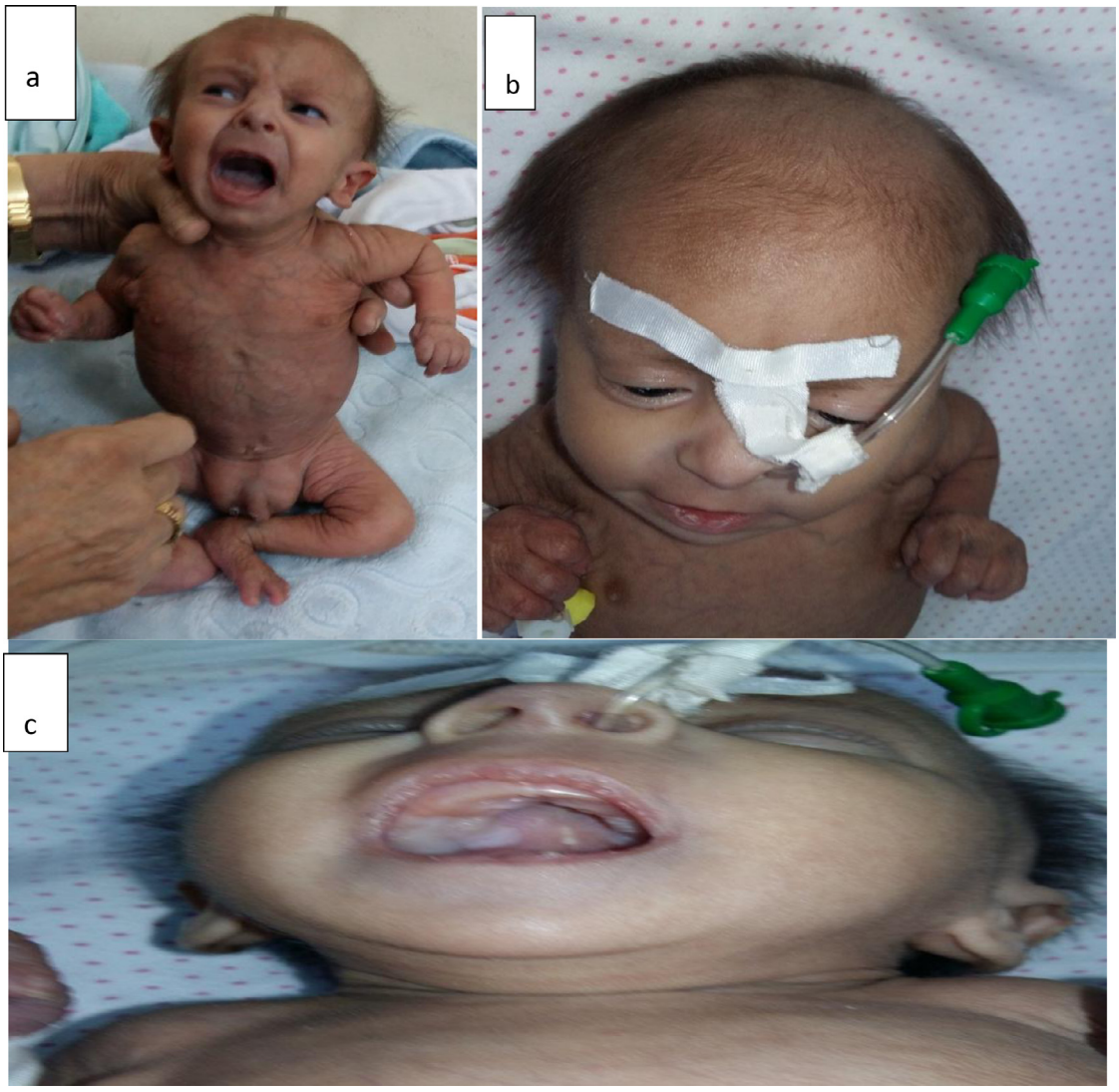

Fig. 1. a) Marked decreased subcutaneous fat, micrognathia and low set ears b) Sparse scalp hair, sparse eyebrows and eyelashes with loss of hair in anterior two thirds of scalp c) High arched palate and short neck.