Case Report

Majewski osteodysplastic primordial dwarfism type II (MOPD-II): A rare case report

Sobhan R[a], Khan HH[b], Pathan MF[c], Afsana F[d], Amin F[e]

Abstract

Majewski osteodysplastic primordial dwarfism type II (MOPD-II) is a rare syndrome characterized by the presence of intrauterine growth restriction, post-natal growth deficiency and microcephaly. Individuals affected by this disease present at an adult height of less than 100 cm, a post-pubertal head circumference of 40 cm or less, mild mental retardation, an outgoing personality and skeletal dysplasia, renal, hematopoietic abnormalities, cerebral vascular anomalies (aneurysm and Moyamoya disease). It is an autosomal recessive syndrome with equal gender occurrence involving the DNA damage-response PCNT gene. Here is an interesting case report of a 15-year-old boy, who presented with growth failure since age of one year, noticed by his parents with history of low birth weight (1.5 kg), delayed developmental milestones, microcephaly, low IQ and difficulty in walking due to short left leg. He had bird like head with beaked nose, crowding of teeth and malocclusion. Complete blood picture and hormonal analysis are within normal range except low growth hormone, typical radiographic features including severe scoliosis and dislocation of hip correlated with MOPD-II. Growth hormone therapy was thought to be ineffective. Genetic counselling is important to prevent the occurrence of MOPD-II.

Key words: *Intrauterine growth retardation, dwarfism, microcephaly, PCNT gene, Majewski osteodysplastic primordial dwarfism, Moyamoya disease.*

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Introduction

Majewski osteodysplastic primordial dwarfism type II (MOPD-II) is a rare autosomal recessively inherited syndrome characterized by severe pre and postnatal growth retardation, short stature, skeletal abnormalities, microcephaly, renal abnormalities and developed aneurysms of the central nervous system arteries and Moyamoya disease.[1,2] MOPD-II is caused by mutations in PCNT (21q22.3).

Author information

a. Dr. Rezwana Sobhan, MD Phase-B, Endocrinology and Metabolism, BIRDEM General Hospital, Dhaka. Bangladesh.

b. Dr. Hafsa Hasan Khan, MD Phase-B, Endocrinology and Metabolism, BIRDEM General Hospital, Dhaka. Bangladesh.

c. Prof. Dr. Md. Faruque Pathan, Department of Endocrinology, BIRDEM General Hospital, Dhaka, Bangladesh.

d. Dr. Faria Afsana, Assistant Professor, Department of Endocrinology, BIRDEM General Hospital, Dhaka, Bangladesh.

e. Dr. Feroz Amin, Associate Professor, Department of Endocrinology, BIRDEM General Hospital, Dhaka, Bangladesh.

Address of correspondence: Dr. Rezwana Sobhan, MD phase-B, Endocrinology and Metabolism, BIRDEM General Hospital, Dhaka. Email: irem89dmc@gmail.com

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Case report

A 15-year-old male student, studying in a school for special children, presented with growth failure noticed by his parents since the age of one year and difficulty in walking due to asymmetrical limbs since the age of 2 years.

He is the first issue of consanguineous marriage, delivered vaginally as a preterm, low birth weight baby with uneventful gestational period but had history of perinatal feeding complications and required admission in neonatal intensive care unit. But his developmental milestones and intellectual development was slightly delayed. He started developing secondary sexual characteristics from the age of 12 years. His parents and siblings are of normal height and there was no family history of short stature or delayed puberty.

On examination, he had disproportionate short stature with short limbs, height was 96 cm (below the 3rd centile), upper segment was shorter than lower segment due to scoliosis. Weight 11 kg, BMI 11.9 kg/m^2. He had microcephaly (head circumference 40 cm) with typical facial appearance with beak-like protrusion of the midface (bird headed) (Figure 1) and beaked nose (Figure 2), sparse hair, micrognathia (Figure 3), short tapering proximal phalanges, clinodactyly, left leg was shorter than the right by 2 cm but no bowing of legs and external genitalia examination revealed Tanner stage- IV.

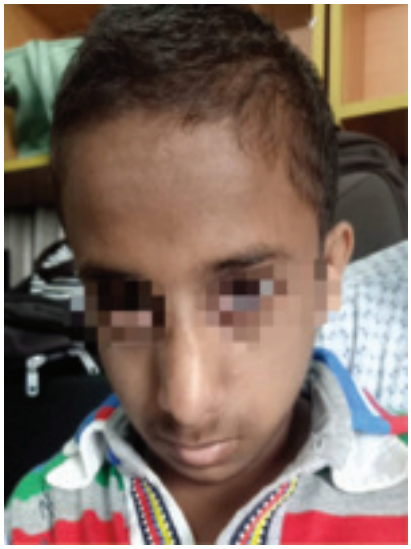

Figure 1 Bird like head and micrognathia

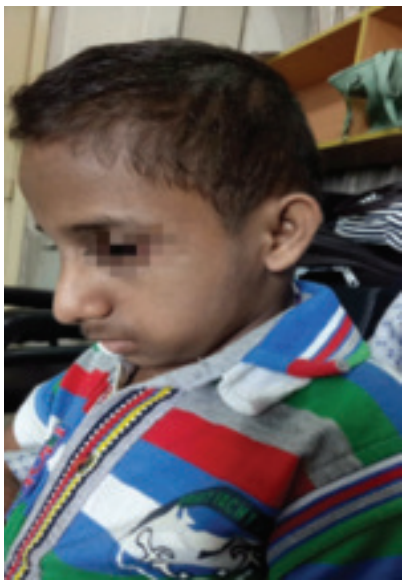

Figure 2 Beaked nose

Complete blood picture and hormonal analysis are within normal range except low growth hormone, typical radiographic features including hypoplastic facial bones and mandibles, clinodactyly, severe scoliosis (Figure 4) and dislocation of hip (Figure 5) correlated with MOPD-II. Growth hormone therapy was thought to be ineffective for this patient as epiphysis were fused rather would be harmful due to severe scoliosis.

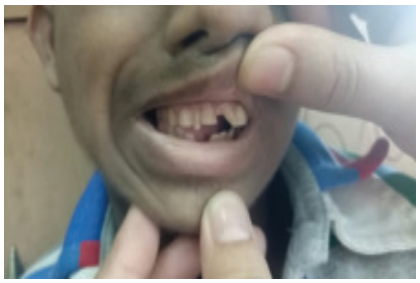

Figure 3 Microstomia and small and crowding of teeth

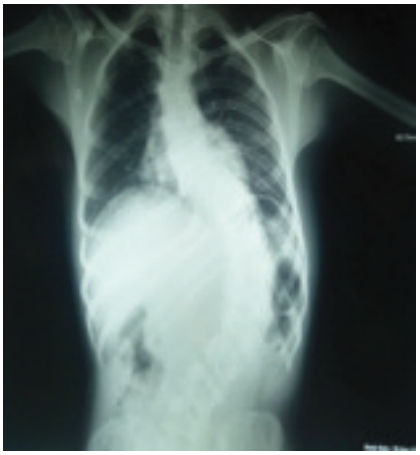

Figure 4 Severe scoliosis

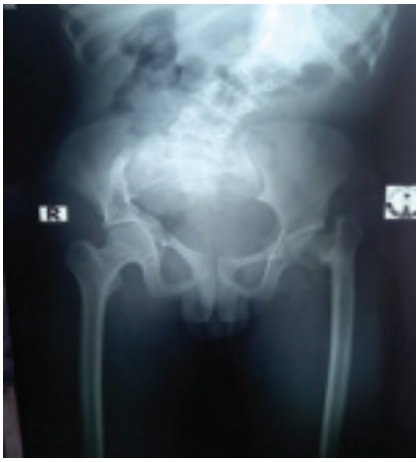

Figure 5 Dislocation of hip

Discussion

MOPD-II is one of the most common forms of primordial dwarfism with more than 150 cases identified worldwide. This condition was initially described by Majewski *et al*. who established the difference between these and Seckel syndrome, a disorder that belongs to the primordial dwarfism group, due to the severity in the growth retardation, the presence of bone abnormalities and mild or absent mental retardation.[2] It occurs equally in male and females and in all ethnic groups. The features of MOPD-II include low birth weight, microcephaly, a prominent nose, dental anomalies including very small, crowding of teeth, scoliosis and kyphosis, shortening of the forearm, dislocation of the hip and bowing of the knees, sparse hair, skin may develop blotchy pigment in sun exposed areas, precocious puberty, farsightedness, renal structural abnormalities in some children, aneurysms which may lead to have stroke-like episodes. These episodes are also common feature of Moyamoya syndrome.[3]

Diagnosis is made by observation of the clinical features, skeletal survey by x-ray, family and medical histories and by molecular testing. The gene for MOPD-II has been identified as *PCNT* on chromosome 21, encoding pericentrin; disruption of pericentrin is thought to cause mitotic spindle defects and impaired cell proliferation and contribute to the development of a wide variety of pathological conditions, including cancer and diabetes.[4] Aneurisms can be diagnosed by a cerebral angiography.

There is no cure for MOPD-II and treatment is designed to help reduce the symptoms. Many children with MOPD-II have feeding problems and can benefit from help such as frequent feeding with small amounts and in some cases, with naso-gastric feeding. Growth hormone does not help.

Monitoring for intracranial vascular abnormalities with brain magnetic resonance angiography is recommended at diagnosis and every 12 to 18 months. Yearly screening for signs of insulin resistance including a lipid profile should be performed, as well as monitoring for anemia, platelet counts and hip and spine anomalies, the eyes for the development of farsightedness, renal anomalies, regular dental monitoring, use of sunblock because of skin fragility. In some cases, orthopedic or psychological support may also be necessary. Genetic counselling is important to prevent the occurrence of MOPD type-II.

Conflict of interest: Nothing to declare.

References

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