Peer Reviewed **Case Report**

ORAL AND RADIOGRAPHIC FEATURES OF MAJEWSKI OSTEODYSPLASTIC DWARFISM: A CASE REPORT

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ABSTRACT

Majewski osteodyplastic primordial dwarfism type II (MOPD II) is a rare, autosomal recessive disorder characterised by severe intrauterine and postnatal growth retardation. A case report with a distinct rare oral and radiographic feature with MOPD II is presented. It differs from the description of primordial dwarfism with characteristics of small head diameter at birth, which progresses to severe microcephaly and mild mental retardation.

Keywords: Autosomal, panorex, microdontia, small roots

LAY ABSTRACT

A description of a rare genetic disorder in a teenage male is provided with unique radiographic features that differ from what were previously documented in literature.

CASE REPORT

A 16-year-old male patient presented with a complaint of loose, painful teeth. He is the only child of healthy non-consanguineous parents. His mother had a normal pregnancy, but had a caesarean section at 32 weeks. He was on treatment for hypothyroidism and has been taking growth hormones from the age of four years.

On extra-oral examination, his stature was smaller than expected relative to his age. His complexion was darker than normal. He had a prominent nose, nasal bridge and a high-pitched voice. He seemed shy at first until he was comfortable after which he interacted well with the healthcare professionals that examined him.

Intra-oral examination of the soft tissue revealed no abnormality. His teeth were however smaller than one would expect for his age. Some teeth were mobile and teeth 55 and 65 were retained. Generalised caries was noted. He was referred by the department of human genetics and underwent testing which confirmed that he has Majewski osteodysplastic primordial dwarfism (MOPD) type II on molecular diagnosis.

Radiographic examinations were performed to assist with further management of his disorder. A lateral cephalometric projection of the face (Figure 1) showed proportional growth of both jaws with a class 1 occlusion. A widening of the sella turcica was noted. A calcified deposit was identified in the region of the sella turcica. The cervical vertebral bodies maturation stage of C4, C5, C6 and C7 were not typical for his age. A panorex radiograph of the maxilla and

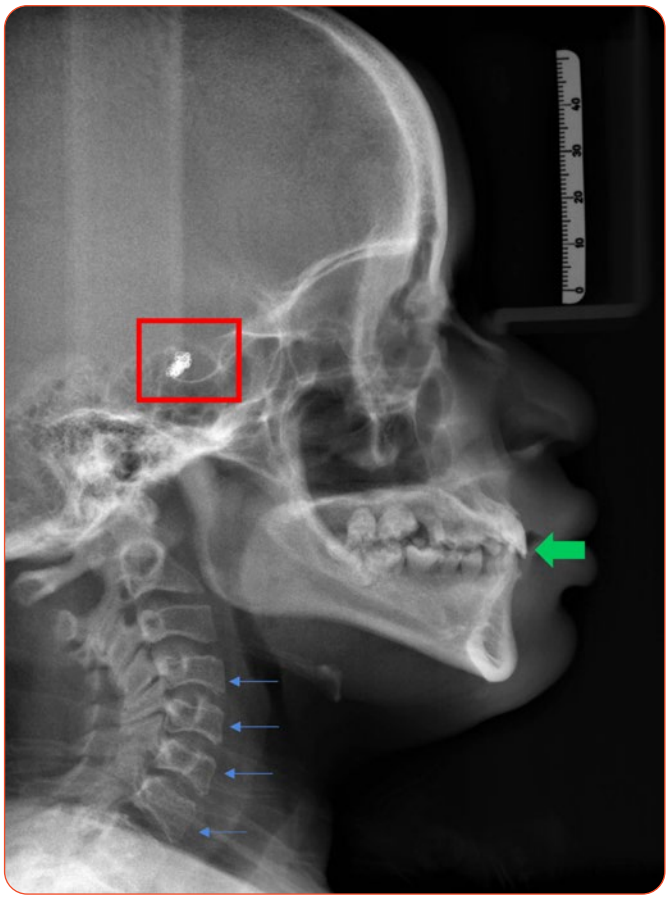

Figure 1. Lateral cephalometric projection of the skull and face shows. Class 1 occlusion (green arrow). Widened sella turcica and calcified deposit (red box). Concave cervical bodies (blue arrows).

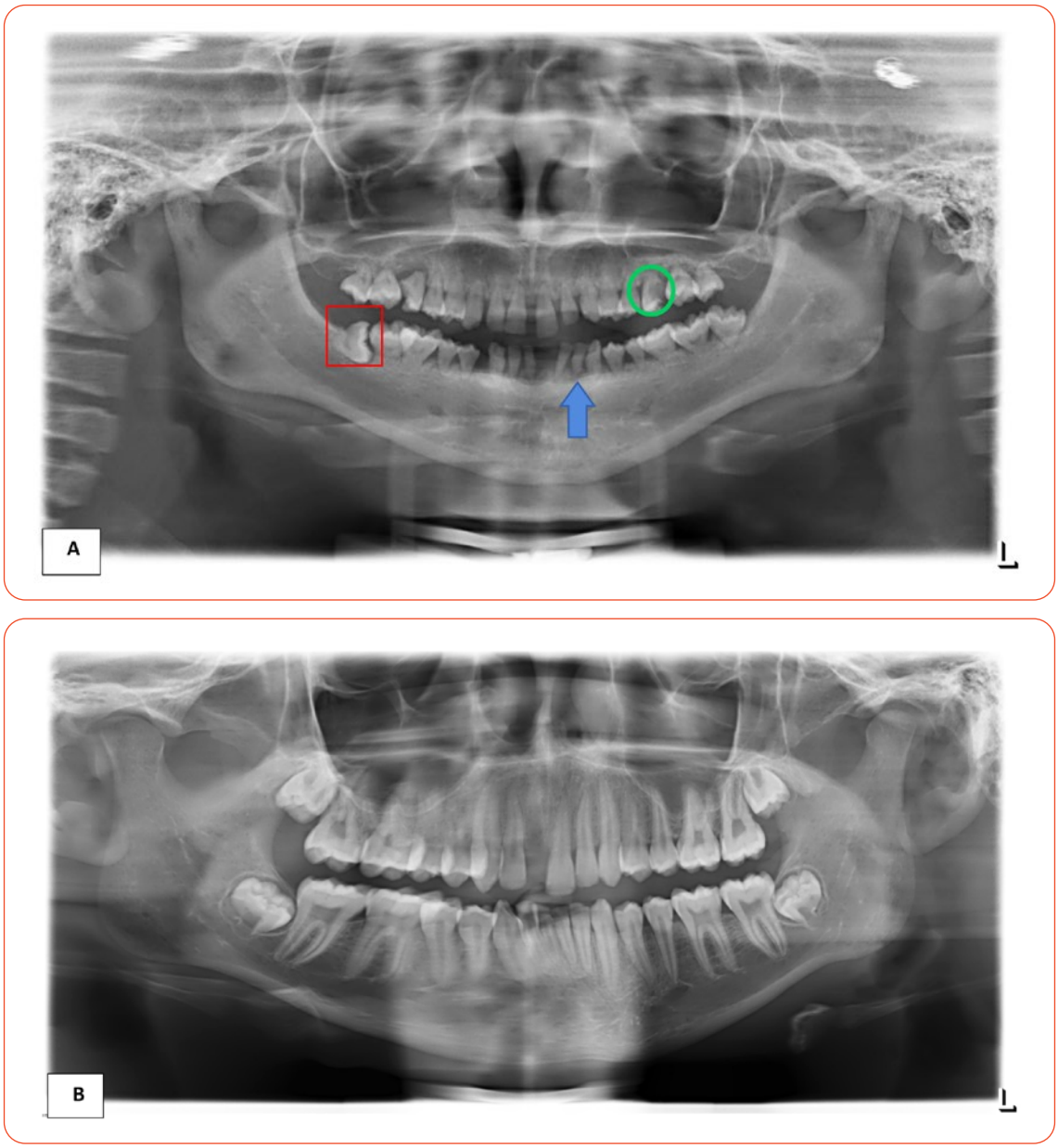

Figure 2. Figure A demonstrates a smaller maxilla and mandible compared to that of a normal 16-year-old in Figure B. Microdontia teeth with short roots of permanent dentition were noted with an example pointed out (blue arrow). There is horizontal impaction of the right lower third wisdom teeth (red box). The upper right molar demonstrates a carious lesion with periapical radiolucency (green circle).

mandible was performed (Figure 2a). The maxilla and mandible were smaller than expected for his age in comparison to a panorex of a normal 16-year-old. Permanent dentition showed microdontia with small roots (Figure 2a). There was horizontal impaction of the right lower third wisdom tooth. The upper first molars had a huge carious lesion with periapical radiolucency.

A dorsi-palmer projection of the hand (Figure 3) demonstrated ossification of a sesamoid bone in the thumb region, which is prevalent in post-puberty. The radiolucencies in the diaphysis-epiphysis of metacarpals are atypical. The fusions of the bases of the 2nd to 5th phalanges and distal ulnar epiphysis, as well as the capping noted at the distal radial epiphysis, are atypical. A full-mouth series was performed. The periapical radiographs are shown in Figures 4a and b. A combination of microdontia, shortness of roots and relatively thick lamina dura were demonstrated in some areas.

DISCUSSION

Majewski osteodyplastic primordial dwarfism type II (MOPD II) is a rare, autosomal recessive disorder caused by mutations in the pericentrin gene in some individuals.[1] It belongs to a heterogeneous group of disorders of primordial dwarfism characterised by severe intrauterine and postnatal growth retardation.[2] Features of this syndrome include severe proportionate intrauterine growth retardation

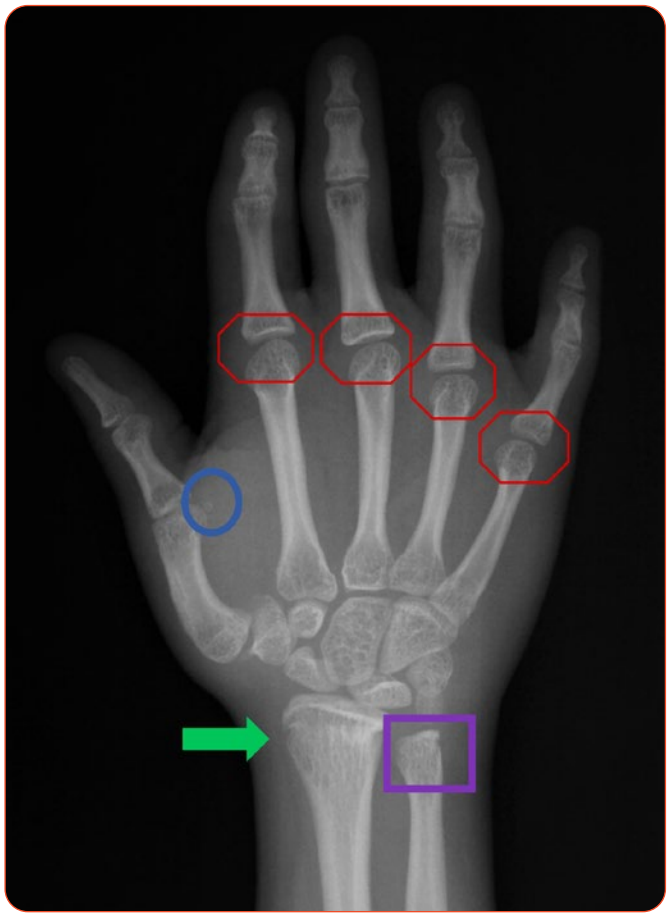

Figure 3. Ossification of sesamoid bone of the thumb (blue circle). Fusion of the 2nd to 5th phalange bases (red octagons). Capping of the distal radial epiphysis (green arrow). Fusion of the distal ulnar epiphysis (purple box).

(IUGR), poor postnatal growth with adult stature of approximately 100 cm, microcephaly which becomes progressively disproportionate, skeletal dysplasia, and characteristic facial features, such as a prominent nose and small jaw.[1,3] The patient in this case report presented with short stature and small body frame. Galasso described this characteristic as small frame.[2] Short stature is believed to be caused by multiple molecular defects, including intracellular signaling, extracellular matrix, and paracrine and endocrine regulation.[4] The approach in the past to short stature primarily focused on clinical manifestations: for example, primordial dwarfism, syndromic short stature or skeletal dysplasia to categorise them by similar clinical features. Currently, the combination of the clinical approach and improved genetic diagnosis are advancing our understanding of congenital growth disorders and have helped further expand the understanding of the clinical variability and genetic heterogeneity of short stature syndrome.[4]

Cranial presentation of poor postnatal growth has been described as microcephaly[1] and micrognathia.[2] The cranium of the patient in this case report had proportional microcephaly. It is defined as a head circumference more than two standard deviations (SD) below the mean for gender and age, and it affects 2% of the population. The patient in this case report did not have micrognathia: his mandible demonstrated proportional growth (Figure 1).[5] Widening of his sella turcica was noted. This feature is described by Terlemez et al.[6]

Literature highlights that a patient with primordial dwarfism has a small head diameter at birth, which progresses to severe microcephaly and mild mental retardation.[4] However,

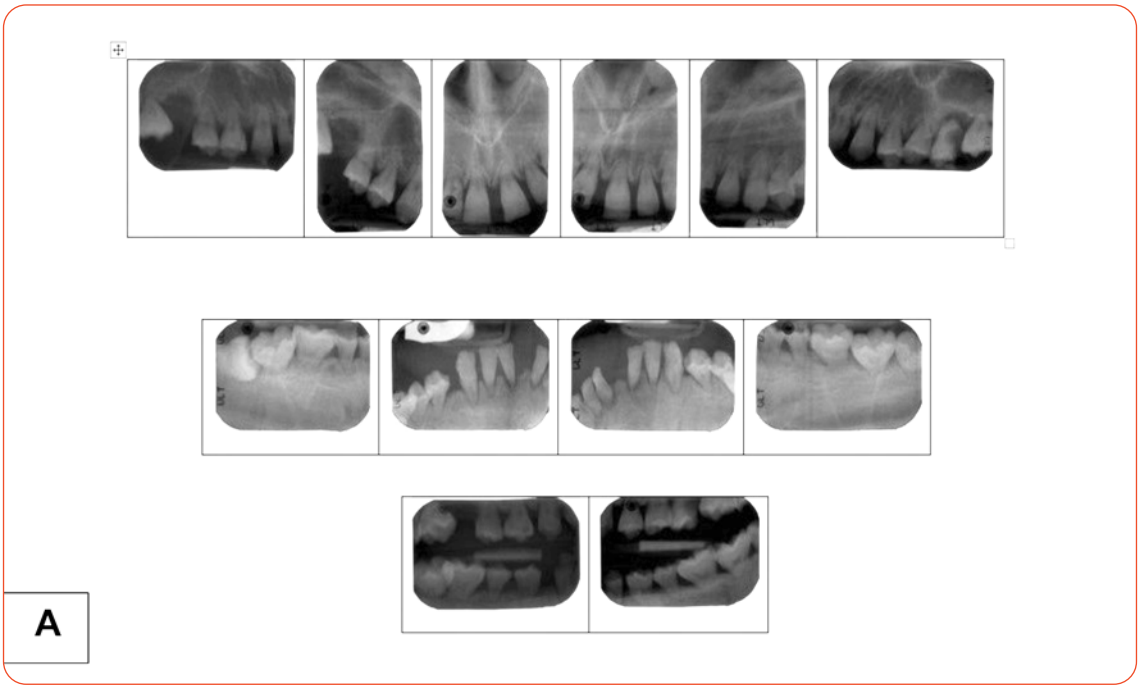

Figure 4a. A full mouth series shows a combination of microdontia and shortness of roots.

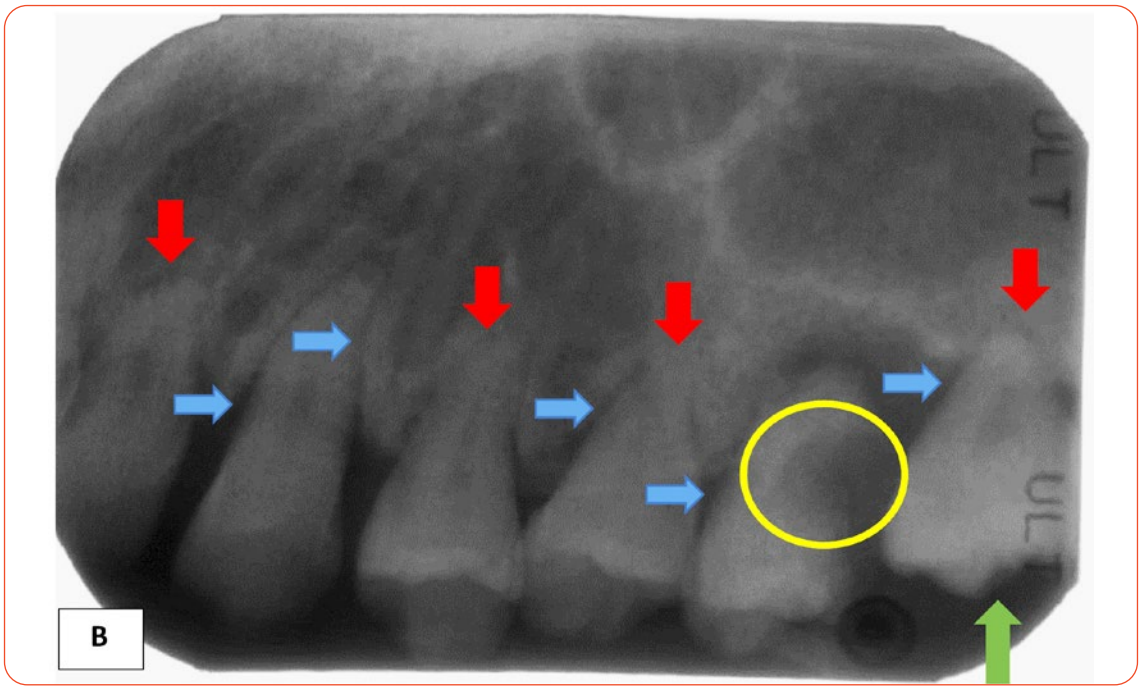

Figure 4b. Periapical radiography of the upper left maxilla i.e. premolar and molar, indicating thick lamina (blue arrows), short roots (red arrows), microdontia (i.e., small tooth) (green arrow) and a carious lesion with periapical radiolucency (yellow circle).

the intellectual capacity of the patient in this case report was above average, based on his performance at school. Most individuals affected by MOPD II are outgoing, talkative and have good social skills.[7] This was the case for this patient and it could also be attributed to relatively good postnatal cranial growth. Distinctive facial features with mild, down slanting palpebral fissures, prominent nose, hypoplasia of the alae nasi, microdontia, micrognathia, and low-set, dysplastic ears have been associated with MOPD II.[2] According to Lui et al.[8] severe micrognathia and mandibular hypoplasia can be diagnosed prenatally by ultrasound. Low-set ears are usually associated with micrognathia involving the mandible.[2] Since there was a proportional postnatal growth of the cranium, the appearance of low-set ear was not noted in the patient in our case.[6] The combination of hypoplastic alveolar processes, microdontia, short-rooted incisors, and rootless molar teeth has been described in MOPD.[4,6] Terlemez et al.[6] reported a female patient with MOPD II with microdontia in the primary dentition.[6] According to Kantaputra et al.,[9] dental anomalies can be striking and present as severe microdontia, opalescent and rootless molars, and an unerupted tooth.[9] The alveolar process can also be severely hypoplastic and mandibular premolars are unusually small and malformed, comprising many cusps.[9] The patient in this case report had microdontia, short roots and mobile teeth in his permanent dentition. This suggests that both primary and secondary dentition were affected by microdontia. Since both dentitions were affected and this implies that the genetic defect occurs before odontogenesis; before the four weeks of embryonic life.[7]

Farman[10] found when determining dental maturity from panorex radiographs that very low birthweight in children tends to result in a delay in dental maturation compared to normal birthweight children. Dental development can also be advanced in a patient with systemic syndromes; the administration of growth hormones had no effect.[10] In clinical practice when determination of skeletal age is done, skeletal development is an important maturity indicator and is helpful in the diagnosis of disorders of growth and development. Skeletal maturation in the vertebral column and hand and wrist regions are reliable anatomical sites for maturity determination.[10] There are several indications of atypical and premature maturity in the patient discussed in our case. The dorsi-palmer hand projection demonstrated premature ossification of the sesamoid bone in the thumb region, fusion of the distal ulnar epiphysis and capping of the distal radial epiphysis (Figure 3). The panorex radiograph showed a fully erupted wisdom tooth, which is another indication of early maturity (Figure 2). Considering the maturity of the cervical vertebrae (Figure 1) the inferior borders of cervical vertebrae 4, 5 and 6 (C4, C5 & C6) were concave and C7 was nearly square-shaped. These features indicate that 5% to 10% adolescent growth can be expected in him relative to his age.[11]

CONCLUSION

The patient in this case presented with most of the symptoms associated with MOPD II. However, the symptoms relative to the clinical presentation suggest that his symptoms are mild. Our case report contributes to the literature as it showed severe microdontia, normal intellectual capacity and premature eruption of a wisdom tooth.

CONFLICT OF INTEREST

None to declare.

ETHICS APPROVAL

Informed consent to use records and to publish was obtained from the patient. Ethics approval was granted by Human Research Ethics Committee of Witwatersrand [M1910101].

CONTRIBUTIONS OF AUTHORS

NJM and MM both contributed to the conceptualisation and writing of this paper.

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