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Dental characteristics in Pallister-Killian Syndrome using Cone Beam Computed Tomography: Illustrated case report

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Abstract

Pallister-Killian Syndrome (PKS) is a rare genetic disorder characterized by the mosaic presence of a supernumerary isochromosome consisting of two short arms of chromosome 12, leading to a variety of complications, including those related to oral and dental health. This case report marks the first case report on the dental characteristics of a patient with PKS using cone beam computed tomography (CBCT). A 17-year-old female with PKS was reported exhibiting bilateral maxillary sinus hypoplasia, along with taurodontism in teeth n°15 and n°16, and an unusual "crayon-like" morphology in teeth n°14, n°15 and n°25. Moreover, hypoplasia of the sphenoidal sinuses, accompanied by left sphenoidal sinusitis, was noted.
A multidisciplinary dental approach is advocated for managing individuals with PKS. Further research incorporating a larger sample is critical to comprehensively assessing the dental features of PKS using dental radiography. Additionally, the development of an open-source CBCT database cataloging dental characteristics for rare dental conditions, including PKS, may be a suitable tool to access to reference images and to share information on orphan diseases with dental implications.

Keywords: Pallister-Killian syndrome, tetrasomy 12p, CBCT, case report

Introduction

Pallister-Killian Syndrome (PKS) is a genetic disorder characterized by its rarity and complexity, with an estimated prevalence of 1 in 20,000 live births [1, 2], exhibiting a moderate female predominance [3]. This condition is caused by a chromosomal anomaly involving a mosaic presence of a supernumerary isochromosome made up of two short arms of chromosome 12, also known as isochromosome 12p [4]. This mosaic pattern means that the chromosomal anomaly is not uniformly present across all cells, leading to a wide range of clinical manifestations without a clear correlation between genotype and phenotype. The syndrome was first brought to light in 1977, marking the beginning of its understanding within the medical community [5]. The PKS can be diagnosed by fluorescence in situ hybridization (FISH) using chromosome 12p-targeted DNA probes analyzing buccal mucosa specimens [2, 5, 6].

PKS exhibits a diverse spectrum of clinical features, encompassing both physical and neurological aspects. Patients often present with distinctive craniofacial dysmorphisms, including but not limited to sparse hair, broad nasal bridges, and unique facial structures [1, 5]. Beyond external appearance, the syndrome affects various systems, leading to intellectual disabilities, hypotonia, and a range of congenital anomalies such as diaphragmatic hernias and heart defects [1, 5, 7]. The complexity extends to metabolic and physiological challenges, including skin pigmentation anomalies and potential sensory impairments [5, 7]. Arghir et al. [8] documented three cases of PKS involving two female and one male subjects. They highlighted several infrequently observed characteristics, including pronounced hypertrichosis on the forehead and ears observable in the initial months post-birth, a distinct ocular condition characterized by atypical iris pigmentation and conical pupil shape, anomalies in connective tissue, recurrent infection episodes, and signs of autonomic nervous system irregularities [8].

Despite its complex presentation, the dental implications of PKS are not extensively documented, with limited cases reporting unique oral characteristics. Bagattoni et al. [9] conducted a clinical dental assessment on a cohort of 21 Caucasian individuals diagnosed with PKS. In their findings, 57% of the individuals exhibited an atypical dental morphology, predominantly characterized by anodontia, with the first permanent molars most frequently absent [9]. The severity of both gingivitis and dental caries exhibited a positive correlation with age [9]. Regarding the occlusion, a high-arched palate and mandibular prognathism were observed, leading to a Class

III malocclusion with both anterior open bites and unilateral or bilateral posterior crossbites [9]. Other oral characteristics have been reported in the literature, including delayed tooth eruption, specific lip formations often referred to as "Pallister lip," and other structural dental anomalies [10]. Treatment for PKS remains supportive and symptom-based, with no cure currently available. Management strategies focus on addressing individual symptoms and improving the quality of life for those affected.

This case report aims to describe the dental characteristics of PKS on cone beam computed tomography (CBCT). To our knowledge, this report is the first to detail the dental characteristics of an individual diagnosed with PKS on CBCT.

Case report

A 17-year-old patient diagnosed with PKS was referred to our clinic for a detailed dental and craniofacial evaluation using CBCT. The assessment focused on the position of teeth n°13, 23, and 24. The CBCT scan was performed with a Planmeca Promax 3D MID device (Planmeca Oy, Helsinki, Finland), under the following conditions: X-ray tube voltage at 90 kV, tube current at 10 mA, exposure time of 18.06 seconds, with an image size of 16/6.2 cm and a voxel size of 200 µm. The scan resulted in a Dose Area Product (DAP) of 851 mGy*cm². Additionally, the patient has hearing difficulties, which was noted with the presence of a right auditory apparatus.

The mandible was not evaluated because it was outside the field of view and not captured in the CBCT scan. Concerning the maxilla, a maxillary sinus hypoplasia was observed in both right and left sinuses, accompanied by slight mucosal thickening on both sides (Figure 1).

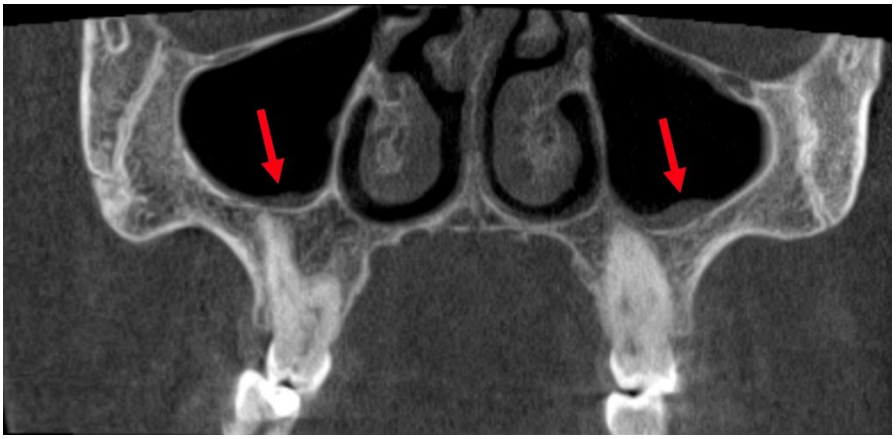

Fig. 1. Planmeca Promax 3D Mid CBCT. Coronal view. Hypodevelopment of right and left maxillary sinus. Red arrows: thickening of maxillary sinus mucosa in the right and left maxillary sinus.

The tooth n°16 exhibited taurodontism [11]. Between teeth n°16 and n°15, either an intraosseous root of a supernumerary tooth or a residual root from the deciduous tooth n°55 was detected (Figure 2A). The tooth n°15 presented with taurodontism [11]. A dental pulp stone was observed in the pulp chamber of the tooth n°17 (Figure 2B).

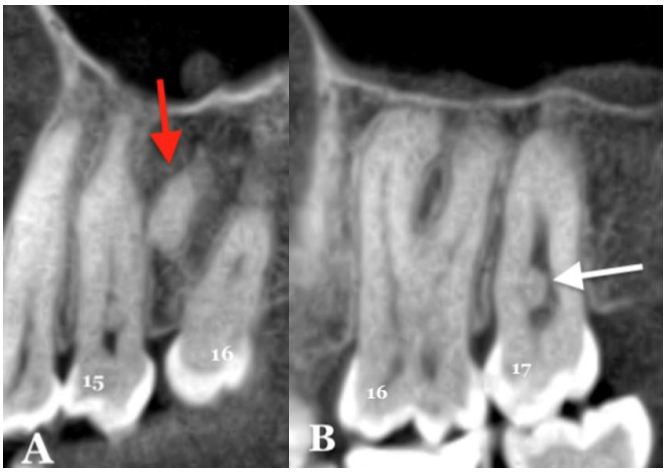

Fig. 2. Planmeca Promax 3D Mid CBCT. Sagittal view of teeth n°15, n°16, and n°17. A. Red arrow indicating the intraosseous root between teeth n°15 and n°16. B. White arrow indicating the pulp stone in tooth n°17. Teeth n°15 and n°16 present with taurodontism.

Teeth n°14, n°15, and n°25 exhibited atypical morphology, characterized by pointed round crowns and elongated roots. Their slender and extended shape resembled crayons, highlighting a distinct "crayon-like" appearance (Figures 3, 4, 6). Measurements from the root apex to the crown tip revealed that tooth n°15 measured 28.36 mm and tooth n°25 measured 28.03 mm.

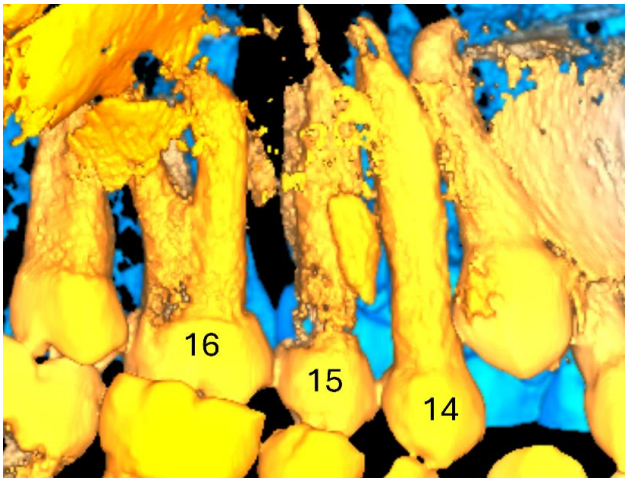

Fig. 3. Planmeca Promax 3D Mid CBCT. Three-dimensional (3D) right lateral view of the upper maxilla. Teeth n°14 and n°15 present with "crayon-like" appearance.

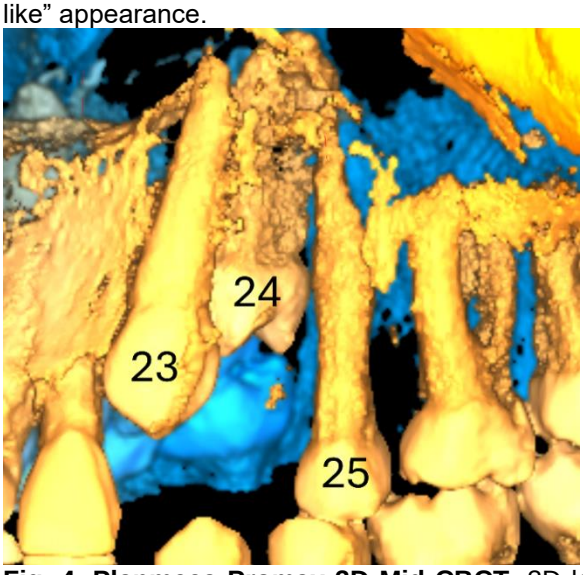

Fig. 4. Planmeca Promax 3D Mid CBCT. 3D left lateral view of the upper maxilla. Tooth n°25 presents with "crayon-like" appearance.

The tooth n°23 was buccally impacted, while the tooth n°24 was palatally impacted. The tooth n°24 was positioned palatally relatively to the tooth n°23, with no signs of external resorption affecting the tooth n°23 (Figures 4, 5).

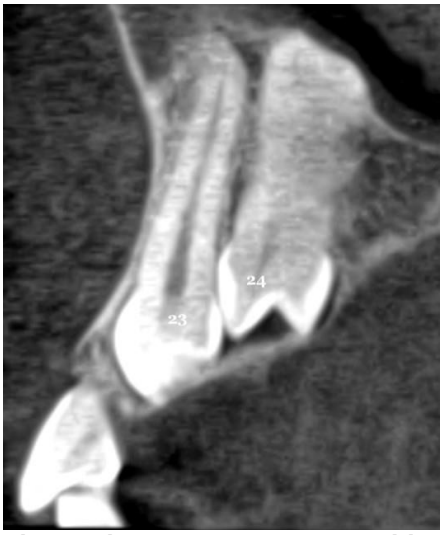

Fig. 5. Planmeca Promax 3D Mid CBCT. Sagittal view. Tooth n°24 palatine to tooth n°23 with close contact but without external resorption of tooth n°23.

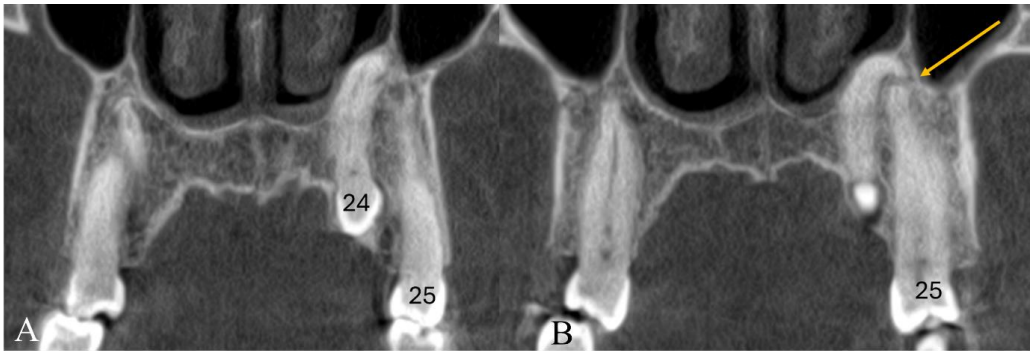

Fig. 6. Planmeca Promax 3D Mid CBCT. Coronal view. A. Tooth n°24 palatine to tooth n°25. The root of tooth n°24 in close contact with left nasal fossa. The tooth n°25 presents with "crayon-like" appearance. B. Dilaceration of the root of tooth n°24 which is above the apex of the tooth n°25 (arrow). The root of tooth n°24 deforms the inferior rim of the left nasal fossa. The tooth n°25 presents with "crayon-like" appearance.

The evaluation of the cervical vertebrae and of the skull base revealed an absence of fusion of posterior arches of C1 (atlas) on the right side of the midline (Figure 7). Furthermore, the skull base exhibited hypoplasia of the sphenoidal sinus, with left sphenoidal sinusitis also being observed (Figure 8).

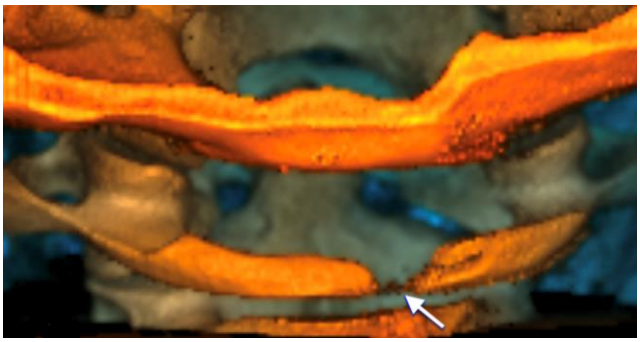

Fig. 7. Planmeca Promax 3D Mid CBCT. 3D reconstruction of cervical vertebrae, posterior view. Absence of fusion of posterior arches of C1 (atlas) on the right side of the midline (arrow).

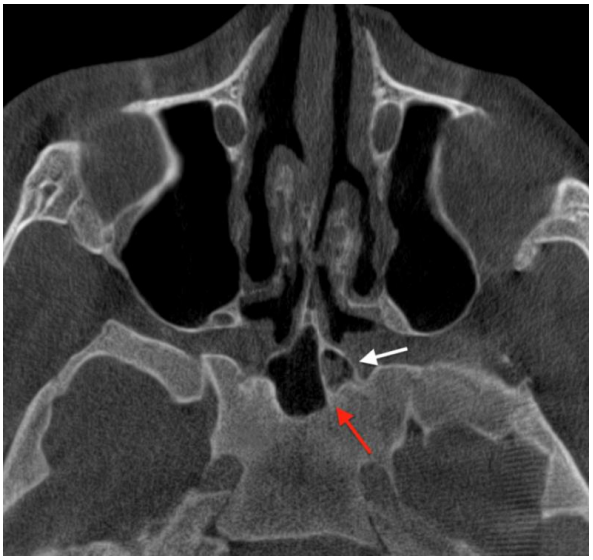

Fig. 8. Planmeca Promax 3D Mid CBCT. Axial view. Hypoplasia of the sphenoidal sinus (red arrow), with presence of left sphenoidal sinusitis (white arrow).

Discussion

PKS is a rare genetic condition, with its dental manifestations sparsely documented within scientific literature. This report aims to fill that gap by looking closely at the dental characteristics of PKS using CBCT.

The reported case revealed bilateral hypoplasia of the maxillary sinuses, which accounted for the patient's facial asymmetry. This observation aligns with the findings by Bagattoni et al. [9], which documented an atypical facial phenotype in patients diagnosed with PKS. Additionally, teeth n°15 and n°16 displayed taurodontism, a condition marked by an enlargement of the pulp chamber, along with the apical displacement of the pulpal floor, making endodontic treatment notably complex [11-13].

Moreover, this report highlights an atypical tooth morphology; teeth n°14, n°15 and n°25 exhibited a "crayon-like" shape with an average length of 28.19 mm, which could present a challenge during endodontic treatment if needed. While hypodontia has been reported in several PKS cases [8, 9], it was not observed in the reported case. Instead, the impaction of teeth n°23 and n°24 was noted. Another finding was the hypoplasia of the sphenoidal sinus accompanied by left sphenoidal sinusitis.

This case report enriches the existing clinical data on PKS by providing insights from the CBCT evaluation, particularly highlighting a previously undocumented atypical tooth morphology—characterized by a "crayon-like" appearance on radiographs. Further research involving a larger sample size is essential to evaluate the dental characteristics of patients diagnosed with PKS on dental radiographs.

Finally, development of open-source CBCT database cataloging dental characteristics for rare dental conditions, including PKS, may be a suitable tool to access to reference images and to share information on orphan diseases with dental implications.

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- **Informed consent**: Patient was exempted from the informed consent according to the ethical committee approval.

Authors contribution:

Author	Contributor role
Issa Julien	Conceptualization, Writing original draft preparation
Olszewski Raphael	Conceptualization, Investigation, Data curation, Ressources, Supervision, Validation, Writing review and editing

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