

Case report

Identifying Pyle's metaphyseal dysplasia: An incidental finding of skeletal pathology on a dental checkup

Shruthi Parthasarathy^{a,*} , Amudhan Azhagan^b , Arthisri Anandhi Sekar^{a,c} 
Poongodi V^a , Ram Shankar^a

^aDepartment of Oral Medicine and Radiology Meenakshi Academy of Higher Education and Research (MAHER), Meenakshi Ammal Dental College and Hospital (MADC), Alapakkam main road, Maduravoyal, Chennai, India

^bThangamani multispeciality dental clinic (private practice) A6, paramount park, Vijayanagar, velacherry Chennai, India

^cCentre of Molecular Medicine and Diagnostics (COMManD), Saveetha Dental College and Hospitals, Saveetha Institute of Medical and Technical Sciences, Saveetha University, Chennai, India

Abstract

Pyle's disease, also known as familial metaphyseal dysplasia, is a rare autosomal recessive genetic disorder that primarily affects the skeletal system, particularly in children and adolescents. Furthermore, it is characterized by distinctive radiographic features, including Erlenmeyer flask deformity and metaphyseal widening. Pyle's disease often goes underdiagnosed because of its mild clinical manifestations. This case report details a 15-year-old female patient presenting with retained milk teeth and unerupted permanent teeth, alongside notable skeletal abnormalities, including genu valgum and kyphosis. Clinical and radiographic evaluations revealed considerable dental anomalies, including multiple impacted permanent teeth and mild dysmorphic facial features. Although genetic testing was not performed in this case, a diagnosis of Pyle's disease was made based on the clinical presentation and radiographic findings. This case highlights the importance of recognizing dental manifestations as potential indicators of underlying skeletal dysplasias or other systemic conditions. Prompt diagnosis and early intervention could enhance the quality of life and treatment outcomes in such cases.

Keywords: Erlenmeyer deformity, impacted teeth, metaphyseal dysplasia, orthopedics, Pyle's dysplasia.

Pyle's disease, also known as familial metaphyseal dysplasia, is an exceedingly rare autosomal recessive genetic disorder that primarily affects the skeletal system, particularly in children and adolescents.⁽¹⁾ This condition was first described by Pyle in 1931 and is characterized by distinctive radiographic features, including Erlenmeyer flask deformity of the long bones and metaphyseal widening, which can result in substantial skeletal abnormalities.⁽²⁾ Despite its potential for severe complications, Pyle's disease often presents with mild clinical symptoms, resulting in frequent underdiagnosis or misdiagnosis.⁽³⁾ The disorder is commonly associated with consanguineous

marriages, underscoring the importance of genetic factors in its etiology. Clinical manifestations may include skeletal deformities such as genu valgum, kyphosis, and mild dysmorphic facial features.⁽⁴⁾ In addition, dental anomalies are frequently observed, including retained deciduous teeth and impacted permanent teeth, which can serve as critical indicators for clinicians to consider underlying skeletal dysplasias.⁽⁵⁾

This case report presents a 15-year-old female patient with notable dental and skeletal abnormalities that ultimately led to a diagnosis of Pyle's disease. Informed consent was obtained from the patient for the inclusion of clinical images in the manuscript. By documenting this case, we aim to highlight the importance of recognizing dental manifestations as potential signs of rare genetic disorders and to enhance awareness among healthcare professionals regarding the implications of such conditions on overall patient management. Our case provides further clinical evidence for this observation, reinforcing the importance of dental imaging as a diagnostic tool.

***Correspondence to:** Shruthi Parthasarathy, Department of Oral Medicine and Radiology Meenakshi Academy of Higher Education and Research (MAHER), Meenakshi Ammal Dental College and Hospital (MADC), Alapakkam main road, Maduravoyal, Chennai, India.

E-mail: putu.aditya.dk@gmail.com

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Only a few case reports of Pyle's disease have been recorded to date (**Table 1**).⁽⁶⁾ It typically presents with very mild clinical symptoms and generally remains underdiagnosed. Moreover, it is also referred to as "familial metaphyseal dysplasia" and has distinct radiographic features with gross metaphyseal undermining that usually involves the long bones and occasionally the cranium.⁽¹⁾

Case report

A 15-year-old female presented to the Department of Oral Medicine and Radiology with a chief complaint of retained milk teeth on the left side and unerupted permanent teeth for more than 10 months after the extraction of milk teeth on the right side. The patient had a history of primary tooth extraction more than one year prior. The patient's parents had a positive history of consanguineous marriage. Both parents were apparently healthy. The patient was the only child and had no siblings, and no other family members across the past two generations had similar manifestations. As the available history did not suggest a clear familial clustering of the condition, a pedigree chart was not made.

Upon clinical examination, the patient's height, weight, and head circumference were average for age. However, the patient reported a positive history of progressive weight loss for the past 5 years. The patient also often felt tired. There was no history of repeated fractures, bone pain, or repeated infections. Patient reported no hearing or vision symptoms. Furthermore, there were no impairments in daily activities aside from the identified skeletal deformities. Other developmental milestones and intelligence were normal. She had prominent genu valgum, mild kyphosis, and arachnodactyly (**Figure 1A and 1B**). A prominent scapula with winging was also evident.

The patient presented with mild dysmorphic facial features. She had a broad nasal root, low-set ears, and mild frontal bossing (**Figure 2A**). Intraoral examination revealed the presence of permanent teeth 11, 12, 16, 17, 21, 22, 26, 27, 31, 32, 36, 37, 41, 42, and 46. No other permanent teeth were clinically visible. Deciduous teeth 63, 64, 65, 73, 74, and 75 were clinically visible (**Figures 2B and 2C**). Dental caries was seen in 75. The patient's oral hygiene was average, and prominent malocclusion was noted.

An orthopantomogram showed fully erupted permanent teeth 11, 12, 16, 17, 21, 22, 26, 27, 31, 32, 36, 37, 41, 42, 46, and deciduous teeth 63, 64, 65, 73, 74, and 75. Permanent teeth 13, 14, 15, 18, 23, 24, 25, 28, 33, 34, 35, 38, 43, 44, 45, 48 were found completely submerged within the bone with incompletely formed roots. Dental caries involving the enamel and dentin

were seen in 75 (**Figure 3A**). Generalized cortical bone thinning was evident in the body of the mandible, ramus of the mandible, and condyle region (**Figure 3A**). Radiographs revealed prominent kyphoscoliosis (**Figure 3B and 3C**).

Metaphyseal widening and flaring were noted in the proximal ends of the femur, fibula, and tibia of both legs. Erlenmeyer flask deformity was observed in the long bones (**Figure 4A, 4B, and 4C**). In addition, there was mild widening of the medial end of the clavicle.

The lateral cephalogram showed small patches of radioopacities scattered throughout the skull, which is suggestive of patchy sclerosis (**Figure 5**).

Blood investigations showed normal serum calcium 9.0 mg/dL (Ref. range: 8.0–10.5 mg/dL), phosphate 3.7 mg/dL (Ref. range: 3.0–4.5 mg/dL), vitamin D₃ 38.0 mg/dL (Ref. range: 30.0–50.0 ng/mL), and magnesium 1.9 mg/dL (Ref. range: 1.7–2.2 mg/dL) levels, and normal thyroid and parathyroid function tests. Routine hematological investigations were also within normal limits.

As the routine blood investigations were normal, thalassemia and lysosomal storage disorders such as Niemann-Pick disease and Gaucher's disease were considered less likely, as these usually present with anemia/thrombocytopenia or bone marrow involvement.

Moreover, as the vitamin D₃ levels, serum calcium, serum phosphate levels, and thyroid and parathyroid function tests were normal, osteopetrosis, rickets, hyperparathyroidism, and hypophosphatemia were also considered less likely.

However, it is paramount that these conditions cannot be definitively excluded without disease-specific investigations/clinical correlation and referral to a general physician. The patient was advised to undergo genetic testing for the SFRP4 gene, but the patient did not consent to it.

Based on all the above investigations, a diagnosis of Pyle's metaphyseal dysplasia was given. The patient was referred to the Department of Orthodontics and Dentofacial Orthopedics for surgical extrusion of all permanent teeth and correction of malocclusion. The nature of the disorder was explained to the patient. Thereafter, she was also referred to the Department of Orthopedics for opinion and further management. Although surgical orthodontic management was discussed as a potential adjunct treatment, after a comprehensive discussion of the risks and benefits, the patient's parents and the patient decided against pursuing any intervention. Therefore, only the clinical features and investigations were described herein.

Table 1. Literature of previously reported cases.

Pt. No	Authors	year	Geographic location	Age of the patient	Clinical / Radiological features	Dental manifestations	Lab investigations	Genetic testing/ family history
1	Bakwin H, <i>et al.</i> ⁽¹¹⁾	1937	Irish American	3 to 9 years follow-up/ male	Knock knees, prominent humerus, limited extension of elbows, weak foot, clubbed fingernails, fusiform expansion of lower ends of femurs and upper ends of tibia, metaphyseal widening of long bones, unusually tall stature (especially lower extremities)	Retained deciduous	Complete blood panel, Dextrose tolerance test, tuberculin test, phosphorous levels, all were within normal limits. ↓ Hb%, ↓RBC count ↑eosinophils	- Sister with similar features
2	Bakwin H, <i>et. al.</i> ⁽¹¹⁾	1937	Irish American	3 to 6 years follow up / female	Knock knees, prominent humerus, limited extension of elbows, weak foot, fusiform expansion of lower ends of femurs and upper ends of tibia, metaphyseal widening of long bones, unusually tall stature (especially lower extremities)		Complete blood panel, calcium, inorganic phosphate, phosphatase, cholesterol, chlorides and sugar were all normal	- Brother with similar features
3.	Komins C, <i>et. al</i> ⁽¹²⁾	1954	England	18 years/ male	Tall stature, long limbs, straightened spine, large skull, hypertelorism, mild genu valgum, hyperostosis of facial bones, obliteration of sinuses and mastoids partially, increased cortication / thickness of skull vault (especially, occipital region and base of skull), large pelvis, expanded clavicles, Erlenmeyer flask deformity, tubular appearances of some phalanges	Mandibular prognathism, speech difficulty, malocclusion, deformed teeth, retained deciduous and dental caries	Norm complete blood panel and chemistry	- Mother, maternal uncle and elder sister had similar features
4.	Komins C, <i>et al.</i> ⁽¹²⁾	1954	England	38 yrs female	Tall stature, large skull, increased cortication / thickness of skull vault (especially, occipital region and base of skull), hyperostosis of facial bones, obliteration of sinuses and mastoids, straightened spine (loss of curve), large pelvis, expanded clavicles, Erlenmeyer flask deformity, tubular appearances of some phalanges	Large mandible, no prognathism	-	- Mother, maternal uncle and younger brother had a few features
5	Ross MW, <i>et al.</i> ⁽¹³⁾	1967	White - Cuba	15 yrs/male	Grotesque facial features and deformity, tall stature, mild genu valgum, palpable heaviness of extremities, large skull, prominent glabella, large zygomatic process, exophthalmos and hypertelorism	Mandibular prognathism		- Younger sister had similar features
6	Ross MW, <i>et al.</i> ⁽¹³⁾	1967	White - Cuba	12 yrs/F	Grotesque facial features and deformity, tall stature, mild genu valgum, palpable heaviness of extremities, large skull, prominent glabella, large zygomatic	Mandibular prognathism		Elder brother had similar features
7	Small PG, <i>et al.</i> ⁽¹⁴⁾	1970	Irish	57 years	Pain and deformity of both lower legs, increased density and thickening of the base of the skull, absence of pneumatisation of paranasal sinuses, sclerosis of petrous ridges, thinning of cortex in long bones, ground glass appearance in trabeculae, bowing of weight bearing bones, tubular metacarpals	Mandibular prognathism, lack of modelling and widening	Mild anaemia was found	
8	Heselson NG, <i>et al.</i> ⁽¹⁵⁾	1979	African	24 years/ female	Genu valgum, widened metaphyses, patchy sclerosis of the skull and its base, partial/ non pneumatisation of paranasal air sinuses, supraorbital prominence, Erlenmeyer flask deformity	Completely edentulous, Mandibular Prognathism		- Similar features in male cousin

Table 1. (Cont.) Literature of previously reported cases

Pt. No	Authors	year	Geographic location	Age of the patient	Clinical / Radiological features	Dental manifestations	Lab investigations	Genetic testing/ family history
9	Heselson NG, <i>et al.</i> ⁽¹⁵⁾	1979	African		Patchy sclerosis of the skull and its base, partial/ non pneumatisation of paranasal air sinuses, supraorbital orimrnc	Multiple caries, malocclusion, mandibular prognathism	-	Family history positive
10	Heselson NG, <i>et al.</i> ⁽¹⁵⁾	1979	African	50 years/ male	Lumbosacral spondylolisthesis, platyspondyly, metaphyseal flaring, skeletal dysplasia, Patchy sclerosis of the skull and its base, partial/ non pneumatisation of paranasal air sinuses Ribs were flared at the costochondral junction	Completely, edentulous, mandibular prognathism	-	- Family history positive
11	Shibuya H, <i>et al.</i> ⁽¹⁶⁾	1982	Tokyo, Japan	61 years/ male	Thick and widened ribs and clavicle, thickened metaphyses of long bones, sclerosis of cranial base, hypopneumatisation of paranasal sinuses,	Mild mandibular prognathism	Routine blood investigations, serum calcium, serum phosphate, alkaline phosphatase level were normal	- Brother and sister have similar clinical features
12	Shibuya H, <i>et al.</i> ⁽¹⁶⁾	1982	Tokyo, Japan	64 years/ male	Thick and widened ribs and clavicle, thickened metaphyses of long bones, sclerosis of cranial base, hypopneumatisation of paranasal sinuses	Mild mandibular prognathism	Routine blood investigations, serum calcium, serum phosphate, alkaline phosphatase level were normal	- Brother and sister have similar clinical features
13	Shibuya H, <i>et al.</i> ⁽¹⁶⁾	1982	Tokyo, Japan	56 years/ female	Thick and widened ribs and clavicle, thickened metaphyses of long bones, sclerosis of cranial base, hypopneumatisation of paranasal sinuses	Mild mandibular prognathism	Routine blood investigations, serum calcium, serum phosphate, alkaline phosphatase level were normal	- Brothers have similar clinical features
14	Shibuya H, <i>et al.</i> ⁽¹⁶⁾	1982	Tokyo, Japan	38 years/ female	Pain in left knee, tall stature, cortical thinning, Erlenmeyer flask deformity, sclerosis of cranial base, hypopneumatisation of paranasal sinuses	Mild mandibular prognathism	Routine blood investigations, serum calcium, serum phosphate, alkaline phosphatase level were normal	-
15	Vohra V, <i>et al.</i> ⁽¹⁷⁾	1987	Tamil nadu, India	29 years/ Male	Knee pain, genu valgum, medial rotation of tibia, long extremities, limited extension of elbows, hypopneumatized mastoid process, thinning of cortex and ground glass trabecular pattern in extremities, widened pubic bones	Supernumerary teeth	Routine blood investigations, serum calcium, serum phosphate, alkaline phosphatase level were normal	-
16	Disler DG, <i>et al.</i> ⁽¹⁸⁾	1993	Boston	24 years/ Male	Metaphyseal widening and cortical thinning of tibia, femur, proximal humeri Underwent radiotherapy	-	-	-
17	Disler DG, <i>et al.</i> ⁽¹⁸⁾	1993	Boston	24 years/ female	Leg pain, hip dislocation, dysplastic bone changes, shallow acetabulum, short femoral neck Underwent radiotherapy	-	-	-
18	Turra S, <i>et al.</i> ⁽¹⁹⁾	2000	Italy	Male	Moderate platyspondyly	-	-	- 1 brother had similar features
19	Turra S, <i>et al.</i> ⁽¹⁹⁾	2000	Italy	Male	Moderate platyspondyly	-	-	- 1 brother had similar features
20	Turra S, <i>et al.</i> ⁽¹⁹⁾	2000	Italy	Male	Severe platyspondyly, biconcave deformity of vertebra, lumbar osteoporotic	-	-	-
21	Turra S, <i>et al.</i> ⁽¹⁹⁾	2000	Italy	Male	Severe platyspondyly, biconcave deformity of vertebra, lumbar osteoporosis	-	-	-

Table 1. (Cont.) Literature of previously reported cases

Pt. No	Authors	year	Geographic location	Age of the patient	Clinical / Radiological features	Dental manifestations	Lab investigations	Genetic testing/ family history
22	Turra S, <i>et al.</i> ⁽¹⁹⁾	2000	Italy	Female	Lumbar osteoporosis	-	-	-
23	Narayanan V, <i>et al.</i> ⁽²⁰⁾	2006	Davangere, Karnataka	17 yrs / female	Metaphyseal widening and thinning of cortices in femur, ground glass opacity of osteoid matrix, giving rise to “Erlenmeyer flask deformity”., diaphyses appeared spaced, the tibia, fibula and humerus also showed bone remodelling defect, widening of medial third of the clavicles, ground-glass opacity of osteoid matrix	Several clinically missing permanent teeth, multiple retained deciduous, crowding, dental caries, generalised rarefaction of jaws, scanty trabeculae, thinning of mandibular cortex, flared neck of the condyle	Routine blood investigations, serum calcium, serum phosphate, alkaline phosphatase level were normal	-
24	Raad MS, <i>et al.</i> ⁽²¹⁾	2008	Cape town, South Africa	24 years/ female	Skeletal dysplasia, Erlenmeyer flask deformity in femur, genu valgum, tall stature, long fingers, sclerosis in the cranial base and vault, ribs and clavicles were expanded, metaphyseal widening	Malaligned teeth	Routine cytogenetic, haematological and biochemical studies were all normal	1 relative has similar features
25	Raad MS, <i>et al.</i> ⁽²¹⁾	2008	Cape town, South Africa	20 years/ female	Modelling defects of the femora	Malaligned teeth	Routine blood investigations, serum calcium, serum phosphate, alkaline phosphatase level were normal	7 relatives had similar features
26	Wonkam <i>et al.</i> ⁽⁴⁾ 2016	2016	Africa	7 years/ male	Facial dysmorphic features, prominent ears, flat frontal area, metaphyseal widening of long bones, Erlenmeyer flask deformity, cortical thinning of long bones, spinal platyspondyly, mild sclerosis of the skull, bossing of forehead.	Mandibular prognathism, retained deciduous, delayed eruption	Routine blood investigations, serum calcium, liver function tests, were normal Karyotyping was normal	-
27	Soares DX, <i>et al.</i> ⁽¹⁰⁾	2016	Brazil	53 years/ female	Genu valgum, joint pain, widening of knees, mild facial dysmorphism, prominent ears, frontal bossing, Erlenmeyer flask deformity, femorotibial metaphyseal flaring, cortical bone thinning, undertubulation of long bones, expanded proximal thirds of clavicle, poor pneumatization of paranasal sinuses and mastoid air cells, arcuate shaped proximal thirds of humerus	Mandibular prognathism, dental caries	3D reconstruction of lower limbs showed Erlenmeyer flask deformity, S shaped tibia was seen T1 weighted MRI was normal	2 siblings had similar manifestations
28	Soares DX, <i>et al.</i> ⁽¹⁰⁾	2016	Brazil	46 years/ female	Genu valgum, joint pain, mild facial dysmorphism, prominent ears, frontal bossing, Erlenmeyer flask deformity, femorotibial metaphyseal flaring, cortical bone thinning, undertubulation of long bones, expanded proximal thirds of clavicle, S shaped tibia	Malocclusion, high arched palate, mandibular prognathism, dental caries	Laboratory tests were normal	2 siblings had similar manifestations
29	Sena J, <i>et al.</i> ⁽²²⁾	2020	Portugal	75 years/ female	Gonalgia, functional limitation of right knee, symmetrical enlargement of knees, genu valgum, wasting of legs, Erlenmeyer flask deformities were evident in femora, in the proximal end, endosteal cortical thickening was seen, lateral bowing of the tibiae, undertubulation of the superior limbs and pelvic changes were noted			

Hb, hemoglobin; MRI, magnetic resonance imaging; WBC, white blood cells.

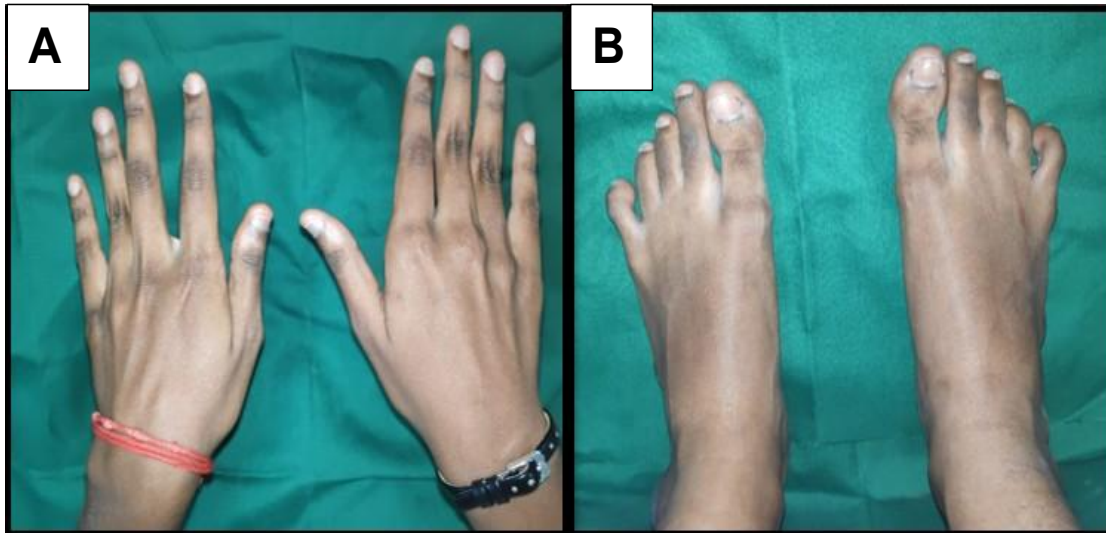


Figure 1. (A) Arachnodactyly of fingers and **(B)** arachnodactyly of toes.

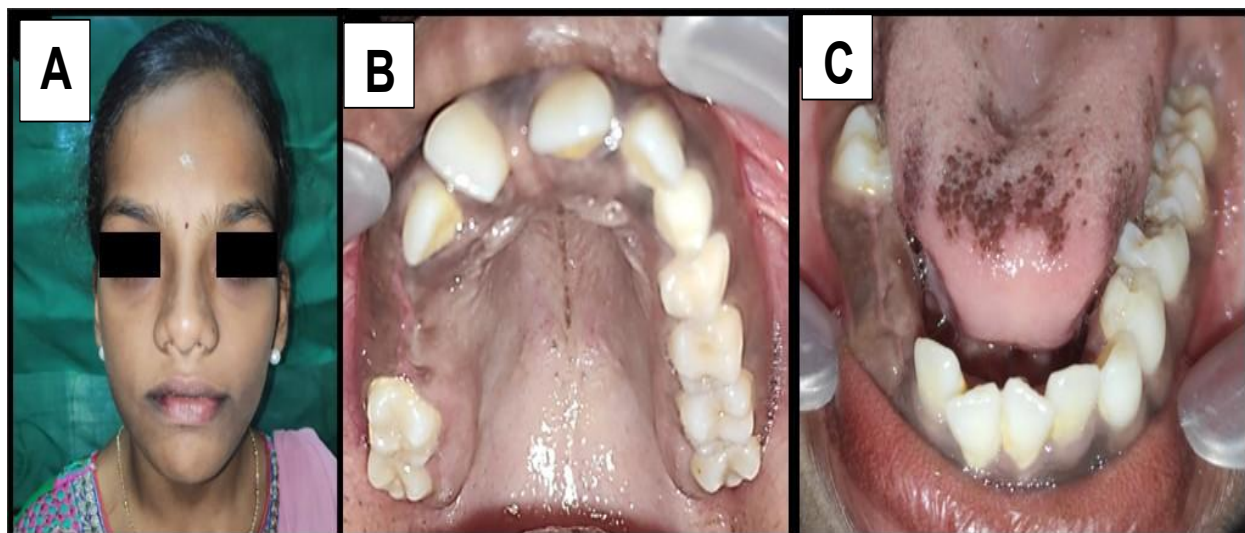


Figure 2. (A) Face showing broad nasal root, low set ears and frontal bossing, **(B)** maxillary arch showing retained deciduous in 63, 64 and 65 & clinically missing 13, 14, 15, 23, 24, and 25, **(C)** mandibular arch showing retained deciduous in 73, 74 and 75 and clinically missing 33, 34, 35, 43, 44, and 45.

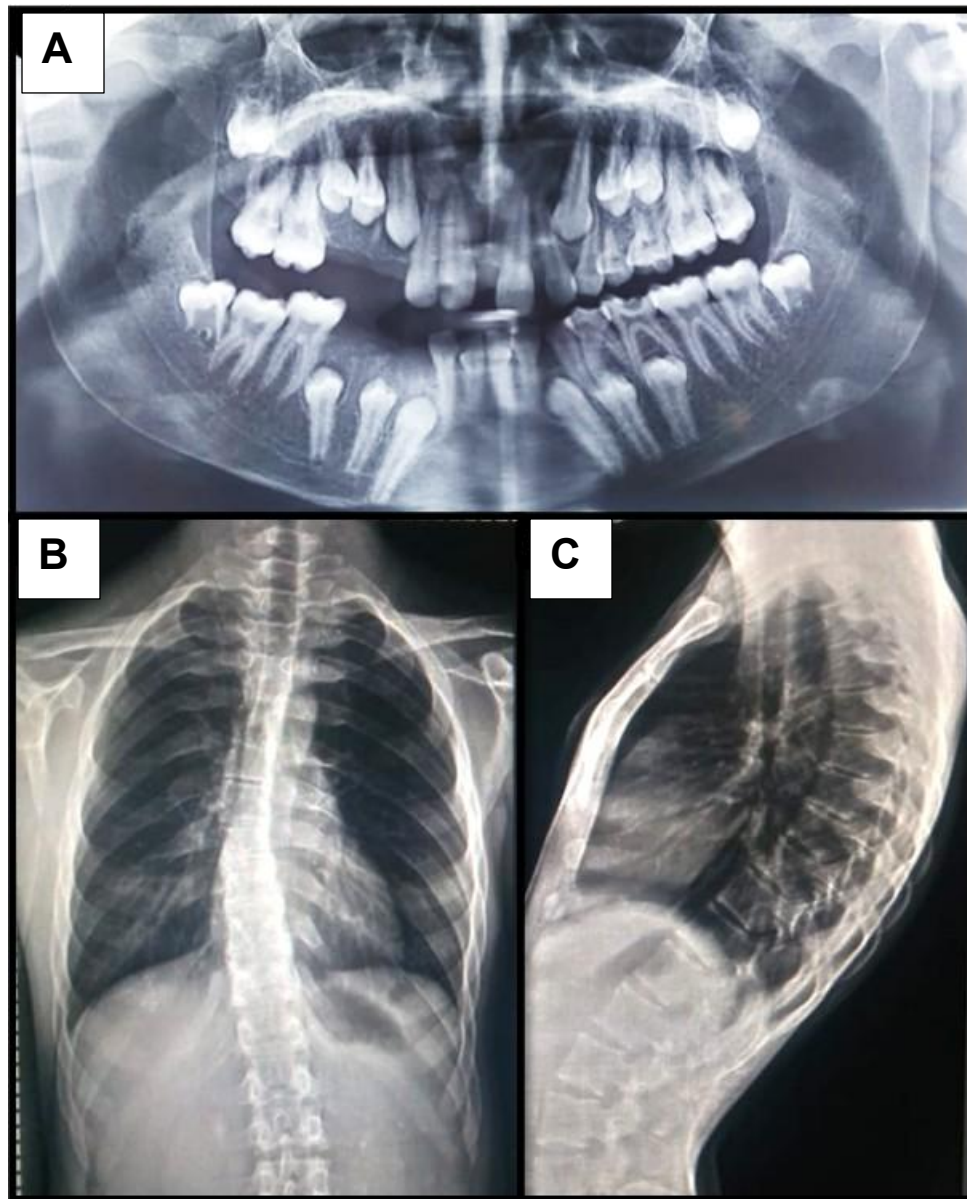


Figure 3. (A) Orthopantomogram showing multiple impacted teeth, (B) scoliosis of the spine, and (C) kyphosis of the spine.

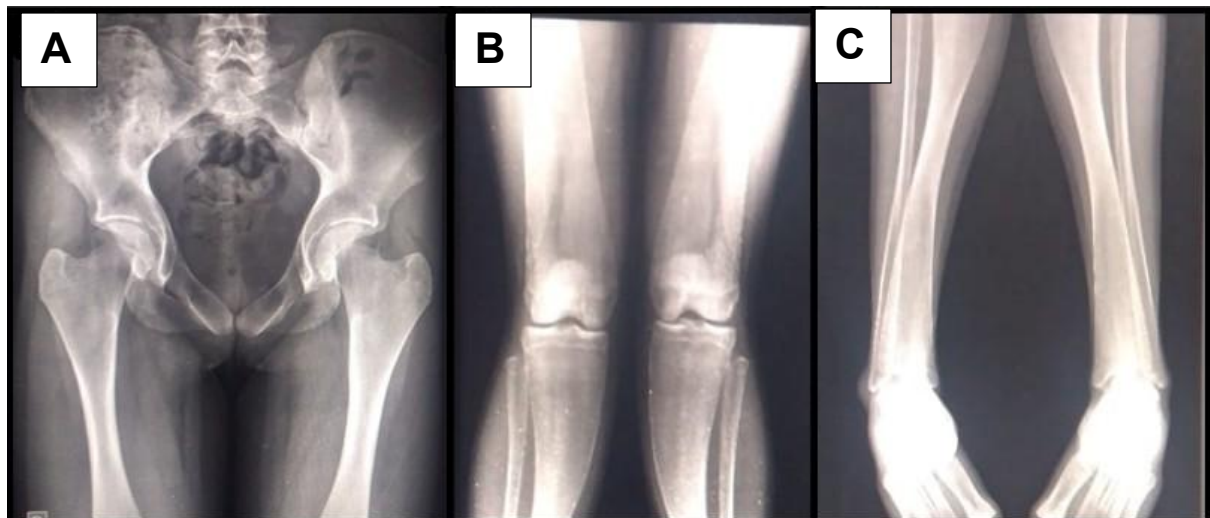


Figure 4. (A) Metaphyseal widening and flaring with Erlenmeyer flask deformity seen in the proximal end of both femurs, (B) metaphyseal widening and flaring with Erlenmeyer flask deformity seen in the distal end of both femurs and proximal end of both tibias, and (C) metaphyseal widening and flaring with Erlenmeyer flask deformity seen in the distal end of both tibias.



Figure 5. Small patches of radiopacities seen all over the skull vault. Mandible is orthognathic with no evident maxillomandibular discrepancies. There were no muscle weakness, no cranial nerve compression, and no history of joint pain.

Discussion

Pyle's metaphyseal dysplasia is an autosomal recessive disorder that can affect siblings and was first documented by Pyle in 1931. It is often associated with the consanguineous marriage of the patient's parents.⁽¹⁾ This disorder is characterized by massive expansion and flattening of the ends of long bones with relative constriction and sclerosis of the diaphysis. Cortical thickening is often evident,⁽⁷⁾ and mild cranial patchy sclerosis may be seen occasionally.⁽¹⁾

The incidental detection of Pyle's disease during dental examinations has been reported previously by Narayanan V, *et al.*⁽²⁰⁾, highlighting the potential for dental evaluations to reveal systemic skeletal anomalies. In the present case, we describe a similar incidental finding and provide an in-depth review of the disease's clinical and radiographic features.

Rather than presenting a novel observation, our objective is to extend the existing literature by providing additional details and a summary of

previously reported cases (**Table 1**), thereby reinforcing the initial discovery and assisting clinicians in managing this rare condition.

When there was cranial involvement, it was initially termed craniometaphyseal dysplasia. However, in 1970, Gorlin *et al.* distinguished craniometaphyseal dysplasia as a separate entity, distinct from Pyle's disease.⁽⁴⁾ Pyle's disease has a distinguished metaphyseal dysplasia and only mild cranial sclerosis, in contrast to craniometaphyseal dysplasia, which shows more severe cranial sclerosis and only mild metaphyseal dysplasia.⁽²⁾

It is one of the causes of an Erlenmeyer flask deformity of the long bones. The flared metaphyses are relatively lucent, and patients are often asymptomatic and identified through routine radiographs.⁽⁸⁾ Genu valgum deformity is a common feature. The ends of the clavicle are widened and may exhibit medial expansion. Spinal involvement may include mild platyspondyly of the vertebra.⁽⁵⁾ Other features include kyphosis, scoliosis, and limited elbow extension.

Although the dental and maxillofacial manifestations of the disease are less severe and less worrisome than the other orthopedic manifestations, they often lead clinicians to suspect a skeletal anomaly and proceed with further investigations. Such was the case detailed above. The most common orodental features of the disease reported in the literature include prolonged retention of deciduous teeth, multiple impacted permanent teeth, malocclusion, mandibular prognathism, and a high incidence of caries.⁽⁴⁾

Although the patient reported a history of progressive weight loss for the past 5 years and frequent tiredness, the initial evaluation did not lead to an underlying systemic cause. However, as previously mentioned, the patient was referred for a thorough systemic examination but was subsequently lost to follow-up. The absence of detailed investigations represents a substantial limitation of this report.

Pyle's disease is a genetic disorder. A previous genetic study of four patients with Pyle's disease demonstrated that the *SFRP4* gene (encoding secreted frizzled-related protein 4) was responsible for these bone changes.⁽⁹⁾ The pathogenesis of the disease remains unclear, but it primarily causes bone resorption failure. According to Pyle, this failure leads to deposition of several layers of undermodeled bone at the metaphyseal segment. Moreover, it is considered that there is abnormal subperiosteal remodeling in the metaphyses. This may be due to chronic hyperemia of the perichondral ring of osteoblasts. This hyperemia may be due to congenital hyperplasia of the perichondral ring arteries.⁽¹⁰⁾

Other disorders with similar manifestations include craniometaphyseal dysplasia, craniodiaphyseal dysplasia, Niemann-Pick disease, Gaucher disease, osteopetrosis, and thalassemia.⁽²⁾

When an Erlenmeyer flask deformity is present, conditions such as craniometaphyseal dysplasia, osteopetrosis, thalassemia, and lysosomal storage disorders must be considered.⁽²⁾ However, the lack of cranial nerve involvement, generalized osteosclerosis, anemia, and other systemic characteristics renders these diagnoses less probable.^(2, 4) Furthermore, the combination of symmetrical metaphyseal widening, mild cranial sclerosis, normal lab results, and serious dental problems, such as delayed eruption, retained deciduous teeth, and multiple impactions, strongly supports the diagnosis of Pyle's disease.^(4, 5)

No intervention is required in most cases. However, bilateral osteotomies can correct progressive genu valgum by temporarily arresting growth in the

growth cartilage on the internal aspect of the tibia and by allowing bone remodeling in the tibial metaphysis.⁽⁷⁾ Patients' bones are extremely fragile and are prone to fractures. Therefore, special care should be taken when performing surgeries or extractions.⁽⁸⁾

Conclusion

This case highlights the crucial role of dental examinations for the early detection of Pyle's disease, a rare genetic disorder that can lead to substantial skeletal deformities if left unrecognized. The patient's presentation of dental anomalies, combined with characteristic radiographic findings, facilitated the timely diagnosis and referral for orthopedic intervention.

While genetic testing was not undertaken for this patient, the clinical features and familial history strongly suggest Pyle's disease. This emphasizes the need for clinicians to maintain a level of suspicion for underlying skeletal conditions in patients presenting with dental issues, particularly in the context of familial patterns and consanguinity. Continued documentation and research into such rare disorders are vital to improving diagnostic accuracy and treatment strategies, ultimately enhancing patient outcomes and quality of life.

Author contributions

SP and AA contributed substantially to the concept and design of this study, acquiring the data, reviewing the literature, and its analysis and interpretation. SP and AA contributed substantially to acquiring the data. SP, AAS, PV, RSS contributed to drafting the manuscript. SP, AAS, PV, RSS edited the manuscript critically for important intellectual content. All authors approved the final version submitted for publication and take responsibility for statements made in the published article

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Conflict of interest statement

All authors have completed and submitted the International Committee of Medical Journal Editors Uniform Disclosure Form for Potential Conflicts of Interest. All authors declare that they have no conflicts of interest.

Data sharing statement

Data generated or analyzed for the present report are included in this published article. Further details are available from the corresponding author on reasonable request after deidentification of the patient whose data are included in the report.

ORCID

Shruthi Parthasarathy  <https://orcid.org/0000-0001-8376-4580>

Amudhan azhagan  <https://orcid.org/0000-0002-7148-1302>

Arthisri Anandhi Sekar  <https://orcid.org/0000-0002-1935-7902>

Poongodi V  <https://orcid.org/0000-0003-1036-0897>

Ram Shankar S  <https://orcid.org/0009-0007-4172-1823>

References

- Beighton P. Pyle disease (metaphyseal dysplasia). *J Med Genet* 1987;24:321–4.
- Faden MA, Krakow D, Ezgu F, Rimoin DL, Lachman RS. The Erlenmeyer flask bone deformity in the skeletal dysplasias. *Am J Med Genet A* 2009;149A:1334–45.
- Chetty M, Roomaney I, Oosterwyk C, Manyisa N, Bope CD, Agenbag G, et al. Craniofacial, dental, and molecular features of Pyle disease in a South African child. *BDJ Open* 2022;8:28.
- Wonkam A, Makubalo N, Roberts T, Chetty M. Pyle metaphyseal dysplasia in an African child: Case report and review of the literature. *SAfri Med J* 2016;106:110.
- Sowińska-Seidler A, Sztromwasser P, Zawadzka K, Sielski D, Bukowska-Olech E, Zawadzki P, et al. The First report of biallelic missense mutations in the SFRP4 gene causing Pyle disease in two siblings. *Front Genet* 2020;11:593407.
- Galada C, Shah H, Shukla A, Girisha KM. A novel sequence variant in SFRP4 causing Pyle disease. *J Hum Genet* 2017;62:575–6.
- Gupta N, Kabra M, Das CJ, Gupta AK. Pyle metaphyseal dysplasia. *Indian Pediatr* 2008;45:323–5.
- Hordyjewska-Kowalczyk E, Wuyts W, Boeckx N, Verdonck A, Hendrickx G, Mortier G. RUNX2-related metaphyseal dysplasia with maxillary hypoplasia: A rare skeletal disorder resembling SFRP4-related Pyle disease. *Clin Genet* 2024;105:434–9.
- Kiper POS, Saito H, Gori F, Unger S, Hesse E, Yamana K, et al. Cortical-Bone Fragility — Insights from sFRP4 deficiency in Pyle’s disease. *N Engl J Med* 2016;374:2553–62.
- Soares DX, Almeida AM, Barreto AR, Alencar E Silva IJ, de Castro JD, Magalhães Pinto FJ, et al. Pyle disease (metaphyseal dysplasia) presenting in two adult sisters. *Radiol Case Rep* 2016;11:405–10.
- Bakwin H, Krida A. Familial metaphysal dysplasia. *Am J Dis Child* 1937;53:1521–7.
- Komins C. Familial metaphyseal dysplasia, Pyle’s disease. *Br J Radiol* 1954;27:670–5.
- Ross MW, Altman DH. Familial metaphyseal dysplasia: review of the clinical and radiologic feature of Pyle’s disease. *Clin Pediatr (Phila)* 1967;6:143–9.
- Small PG, Wallis JJ. Pyle’s disease or craniometaphyseal dysplasia (tarda). *Br J Radiol* 1970;43:811–3.
- Heselson NG, Raad MS, Hamersma H, Cremin BJ, Beighton P. The radiological manifestations of metaphyseal dysplasia (Pyle disease). *Br J Radiol* 1979;52:431–40.
- Shibuya H, Suzuki S, Okuyama T, Yukawa Y. The radiological appearances of familial metaphyseal dysplasia. *Clin Radiol* 1982;33:439–44.
- Vohra V. Pyle’s disease – familial metaphyseal dysplasia – a case report. *Australas Radiol* 1987;31:75–8.
- Disler DG, Alexander AA, Mankin HJ, O’Connell JX, Rosenberg AE, Rosenthal DI. Multicentric fibromatosis with metaphyseal dysplasia. *Radiology* 1993;187:489–92.
- Turra S, Gigante C, Pavanini G, Bardi C. Spinal involvement in Pyle’s disease. *Pediatr Radiol* 2000;30:25–7.
- Narayanan V, Ashok L, Mamatha G, Rajeshwari A, Prasad S. Pyle’s disease: an incidental finding in a routine dental patient. *Dentomaxillofacial Radiol* 2006;35:50–4.
- Raad MS, Beighton P. Autosomal recessive inheritance of metaphyseal dysplasia. *Clin Genet* 1978;14:251–6.
- Sena J, Xavier G, Sampaio J, Nóbrega J. Rare case of Pyle’s metaphyseal dysplasia presenting in an adult. *Int J Case Reports Images* 2020;11:101088Z01JS2020.