INTRODUCTION AND LITERATURE REVIEW

The term Pyknodysostosis (PKD) is derived from Greek, whereby ‘pykons’ means dense, ‘dys’ means defect, and ‘osteosis’ means bone pathology. Maroteaux and Lamy (1962) recognized this condition as an entity in its own right. Previously it was thought to have been a variant of cleidocranial dysostosis, osteopetrosis, fluorosis and heavy metal poisoning. PKD in children is commoner in males than in females, occurring at a ratio of 2:1. It is characterized by short stature (measuring less than 150cm in adulthood), generalized diffuse osteosclerosis, long bone fractures, hypoplastic clavicles and short stubby fingers. Craniofacial features include a large head with frontoparietal bossing, open soft cranial sutures and fontanelles, depressed nasal bridge, beaked nose, obtuse mandibular gonial angle, a high arched grooved palate, maxillary hypoplasia accompanied with relative proptosis, mandibular fractures, osteomyelitis, malpositioned teeth, delayed exfoliation of primary teeth, crossbite, hypercementosis, elongated soft palate precipitating mouth breathing and heavy snoring in addition to periapical cementoma-like lesions in the mandible. Intraoral features include a grooved or furrowed palate, delayed exfoliation of deciduous teeth but timely eruption of the permanent dentition giving rise to crowding, multiple retained teeth, unerupted teeth within follicles and an expanded alveolus. Follicles occasionally get infected leading to chronic suppurrative osteomyelitis.

Parental consanguinity has been identified in more than 30% of the cases as the cause of this autosomal recessive disorder. Karyotyping suggests that the gene which determines PKD is located on the short arm of a small acrocentric chromosome probably G-22. It follows mutations in the CTSK gene situated at 1q 21 that codes for cathepsin k lysosomal cysteine protease that is highly expressed in osteoclasts leading to disturbed bone resorption and remodeling. In this article we present three cases of PKD who presented with diverse
classical clinico-radiologic features.

CASE 1
An 8-months-old boy with delayed milestones presented with complaints of apparent head enlargement since birth; mouth breathing and loud snoring for 3 months. Initially the head enlargement did not disturb the child’s feeding or sleeping. However, the symptoms worsened and the patient was taken to a nearby hospital where a brain CT-scan revealed features of cerebral atrophy with mild hydrocephalus. The parents were advised on shunting but they opted to seek a second opinion at a referral hospital. The child was a 2nd born in a non-consanguineous family with 1st born being a 6-year-old female who was alive and well. The patient had frontoparietal bone bossing (Fig. 1), copious mucous secretion plugging the nostrils and a hypoplastic mandible.

The palate was high-arched and V-shaped with the soft palate elongated almost to the posterior pharyngeal wall. The alveolar ridges were enlarged without deciduous teeth clinically present in the mouth at 8 months (Fig. 2). The maxilla was underdeveloped and the head circumference at 48 cm was normal for the age. The anterior and posterior fontanelles were open and pulsatile. The cranial sutures were open and soft to superficial palpation while the chest was pigeon-shaped and had a rachitic rosary. He had a hepatomegally of 4cm below the costal margin and cardiomegally with a rim of pericardial effusion. The infant had decreased nostrils, a depressed nasal bridge and hypertelorism.

The lateral skull X-ray revealed a very dense calvarium, orbital rims and skull base and; wormian bones were also present in the lambdoid suture. Mandibular angles were obtuse and hypoplastic with evident retained ghost-like teeth (Fig. 1. Inset).

This case also demonstrated dense ribs, hypoplastic lateral ends of the clavicles with extreme ends having been aplastic (Fig. 3). There were dense proximal ends of the humera and a healing fracture of the proximal right humerus. Brain ventricles were enlarged but with an apparent rim of CSF along the frontal poles which ruled out hydrocephalus (Fig. 1 inset: CT SCAN). The patient was remarkably short for his age and had delayed milestones. The diagnosis of PKD in this case was made on the basis of clinical and radiological features.

CASE 2
A 40-year-old man presented with persistent pain in the lower posterior left mandible 6 months after he had had a tooth extraction. The patient was of short stature with pronounced bilateral parietal prominences. On craniofacial inspection, the anterior and posterior fontanelles were clinically unossified and there was frontal bossing with mid and lower facial hypoplasia.
Intraoral examination revealed a non-healed extraction socket in a swollen posterior left mandible. Remarkably, the patient had a high-arched palate with tooth crowding in both arches. A lateral view radiograph of the full cranium revealed the typical osteosclerotic features, an obtuse gonial angle, hypoplastic maxillary bone and underpneumatised maxillary sinuses which were characteristic of PKD (Fig. 4)

CASE 3
A 37–years–old man was referred for the evaluation of a persistent cutaneous draining sinus over his left body of the mandible which had manifested 6 months previously (Fig. 5). Notably, the patient had sustained spontaneous upper and lower limb fractures variously (Fig. 5 inset). Craniofacial clinical inspection revealed patent anterior and posterior fontanelles, a high arched palate and tooth crowding in both arches. An orthopantomograph (Fig. 6) showed generalized tooth hypercementosis with gross features of chronic osteomyelitis in the left body of the mandible characteristic of PKD.

DISCUSSION
Kundu et al and Bathi et al noted that, worldwide PKD has been reported from 9 months to 55 years. Remarkably, we have presented amongst the youngest diagnosed cases, at 8 months with visceral manifestations. The present youngest case had hepatosplenomegaly, cardiomegaly, anaemia, rickets and rachitic rosary as was observed by Kundu et al.

PKD characteristically presents with transverse diaphyseal fractures of the long bones that heal well. This was obvious in case 3 who had lower limb deformity due to repeated fractures with incomplete management but was coincidentally noted in the infant (case 1) as a healing fracture of the right humerus which the mother had no idea had occurred. The infant had not started crawling hence there was a possibility of the fracture having arisen from moderate forces like handling of the
The patient had delayed eruption of primary teeth as was observed by Fleming et al.

The present cases were all males enhancing Wolfgang’s report that PKD was commoner in males than females. However, no parental consanguinity was volunteered in our cases as had been reported by Bathi et al. and Fleming et al. In the developing world, PKD leads to lower limb deformity because of repeated diaphyseal fractures which are hardly treated due to lack of facilities and resources. Since the fractures heal rapidly, patients tend to ignore the deformities despite their severity. Discharging sinus occurs in the jaws because of poor blood supply due to hypercementosis and hyperdense bones. Follicles of impacted teeth may also get infected leading to discharging sinus or chronic suppurative osteomyelitis. However, to date no cystic lesions or bone tumors have been reported in association with follicles of impacted teeth in PKD.

The differential diagnosis of PKD includes cleidocranial dysostosis and osteopetrosis. Notably, cleidocranial dysostosis presents with a normal height, bone texture, gonial angles and the absence of diffuse osteosclerosis. Osteopetrosis may present with stunted growth, a dense skull base, diffuse osteosclerosis, multiple fractures and malunion. The hands, feet, clavicles, gonial angles, maxilla and skull vault are normal. Management of PKD is multidisciplinary: supportive treatment includes the management of anaemia, recurrent infections, failure to thrive, hypocalcaemia, fractures of bones and diverse dental ailments.

ACKNOWLEDGMENT
We are most grateful to the parents of the infant case and the adult patients for consenting to participate in this study. Our sincere gratitude is also extended to the administrations of the Armed Forces Memorial Hospital in Nairobi and the Muhimbili Medical Centre in Dar es Salaam for their permission to execute this study.

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Correspondence Address:
Dr. Jeremiah Moshy,
P. O. Box 65014,
Dar-Es-Salaam, Tanzania.
jeremiahmoshy@yahoo.com

Article Citation:

“Adopt the pace of nature: her secret is patience.”

(Ralph Waldo Emerson)