Constitutional disorders of the bone : rare yet to know : a personal experience and a systematic overview

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Learning objectives

- Remind the classification of this heterogeneous group of disorder.

- Suggest a systematic approach to come up with the right diagnosis.

- Illustrate, through our experience, some of these disorders and their main differential diagnosis.

Background

Constitutional bone disorders are a wide ranged spectrum that includes conditions affecting the growth, the structure or the morphology of the skeleton.

Many classifications have been suggested and none of them offered a systematic approach.

The most common one classify these diseases into two groups: osteochondrodysplasias, which are associated with abnormalities of growth (dysplasias) or texture (osteodystrophies) of bone and / or cartilage; and dysostoses, which are abnormalities of the bone that can be isolated or associated to other deformities.

Dysplasias are often secondary to defects in structural proteins, metabolic processes or in growth plate regulation. Dysostoses arise often from embryonic morphogenic defects.

After the 9th edition of the classification in 2015, we counted overall 436 diseases (was 456) and an increase to 42 groups and 364 genes. It can be explained by the discovery of multiple genectic association.

The diagnosis of congenital bone disorder can be made on the basis of a systematic and elementary approach that integrates clinical, biological and radiological findings.

Findings and procedure details

We retrospectively reviewed the imaging findings of patients diagnosed with congenital bone disorders.

15 patients were enrolled in our study. We counted 9 males and 6 females with an average age of 3.5 years old.

Plain radiography was the main imaging modality and was performed in all our patients.

Findings were as follow: osteoporosis in 4 cases, achondroplasia in 4 cases, Pyle's disease in 2 cases, Leri-weil syndrome in 1 case, osteogenesis imperfecta in 1 case, metaphyseal chondrodysplasia schmid type in 1 case and clinodactyly associated to a Turner syndrome.

Chondrodysplasias are responsible for growth disorders and /or developmental abnormalities of the bone or cartilage. The diagnosis is often made during the early infancy and is suspected in the presence of intrauterine growth restriction or those who present with short stature, especially if there is growth disproportion.

There are lethal neonatal forms namely:

• Achondrogenesis

• Thanatophoric dysplasia

• Platyspondylic lethal skeletal dysplasia

• Atelosteogenesis

• Gracile bone dysplasia

Signs that suggest these forms are:

-Severe micromyelia

-Femur length ratio / abdominal circumference < 0.16

-Chest circumference , growth Percentile < 5 (pulmonary hypoplasia)

-Chest circumference / abdominal circumference <0.8

-Severe polyhydramnios

-Fetal hydrops.

The most frequent dysplasias of prenatal onset are osteogenesis imperfecta type II, thanatophoric dysplasia and achondrogenesis II.

Other most frequent forms of bone dysplasia that evolve through life can be grouped on the bases of the affected site:

• Group I: Epiphyseal dysplasias with or without spinal involvement: Chondrodysplasia punctata, Type II collagenopathies, such as congenital and tardive epondyloepiphyseal dysplasia, Kniest's dysplasia, type II achondrogenesis, spondyloepimetaphyseal dysplasias, multiple epiphyseal dysplasia, pseudoacondroplasia, mucopolysaccharidosis and diastrophic dysplasia.

• Group II: metaphyseal dysplasias: Rhizomelic as in **achondroplasias**; hypochondroplasia and thanatophoric dysplasia and **metaphyseal chondrodysplasia**; or mesomelic or acromelic shortening, such as chondroectodermal dysplasia.

• Group III: Dysplasias with altered bone density: **Osteogenesis imperfecta** and pycnodysostosis.

Osteogenesis imperfacta is a complex group with a clinical and molecular heterogeneity due to lack of type I collagen. Overall there are 4 types:

• Type I: moderate forms with blue sclera

• Type II: lethal forms

• Type III: severe forms (progressive deforming)

• Type IV: similar to type I without blue sclera

Osteogenesis imperfacta affects both bone quality and quantity. The hallmark radiological feature is osteoporosis and fragile bones that can fracture easily (Figure 1).

Achondroplasia is responsible of micromelic dwarfism with rhizomelic shortening (Figure 2-3-4).

It is suspected in the presence of:

- Frontal bulging, mesofacial hypoplasia, narrow magnum foramen

-Short ribs, spinal canal stenosis with decreased interpedicular distance

-In the pelvis the iliac bones have flat acetabular roof and narrow sacrocytic notch.

-Rhizomelic shortened extremities.

Metaphysal chondrodysplasia is characterized by a relatively short stature with shortened extremities and bowed legs

3 types: Schmid type: mild form, due to mutation in type X collagen.

Pena and Vaandrager type: intermediate type

Jansen type: severe form with irregular mineralization in the metaphyses

Metaphysal chondrodysplasia Schmid type: is characterized by a relatively short stature with shortened extremities and bowed legs (genu varum (Figure 4)).

Leri-Weill syndrome is an autosomal dominant dyschondrosteosis characterized by mesomelic shortening of limbs. Patients present with schortened forelegs and forearms. This type of altered development of the lower arm gas often been referred to a Madelung's deformity.

Osteopetrosis (group 26), also called Albers-Schonberg disease, is due to defective osteoclastic activity. It has two varieties, infantile autosomal recessive and benign adult autosomal dominant variety. Infantile or 'malignant' osteopetrosis is a severe form that tends to present early (Figure 7).

Images for this section:

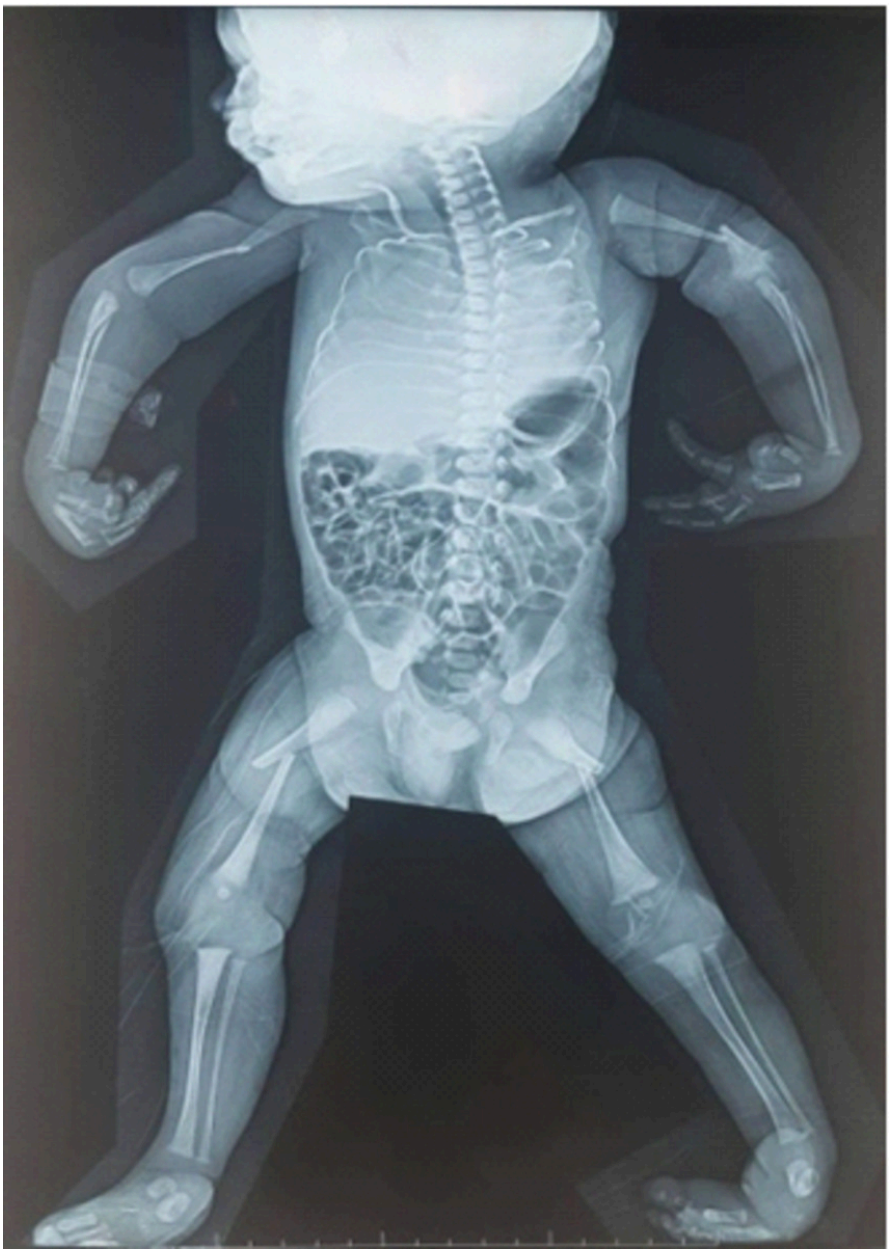

Fig. 1: multiple fractures of the humerus and femurs on the background of abnormally gracile bones: they are bent with thin shafts.

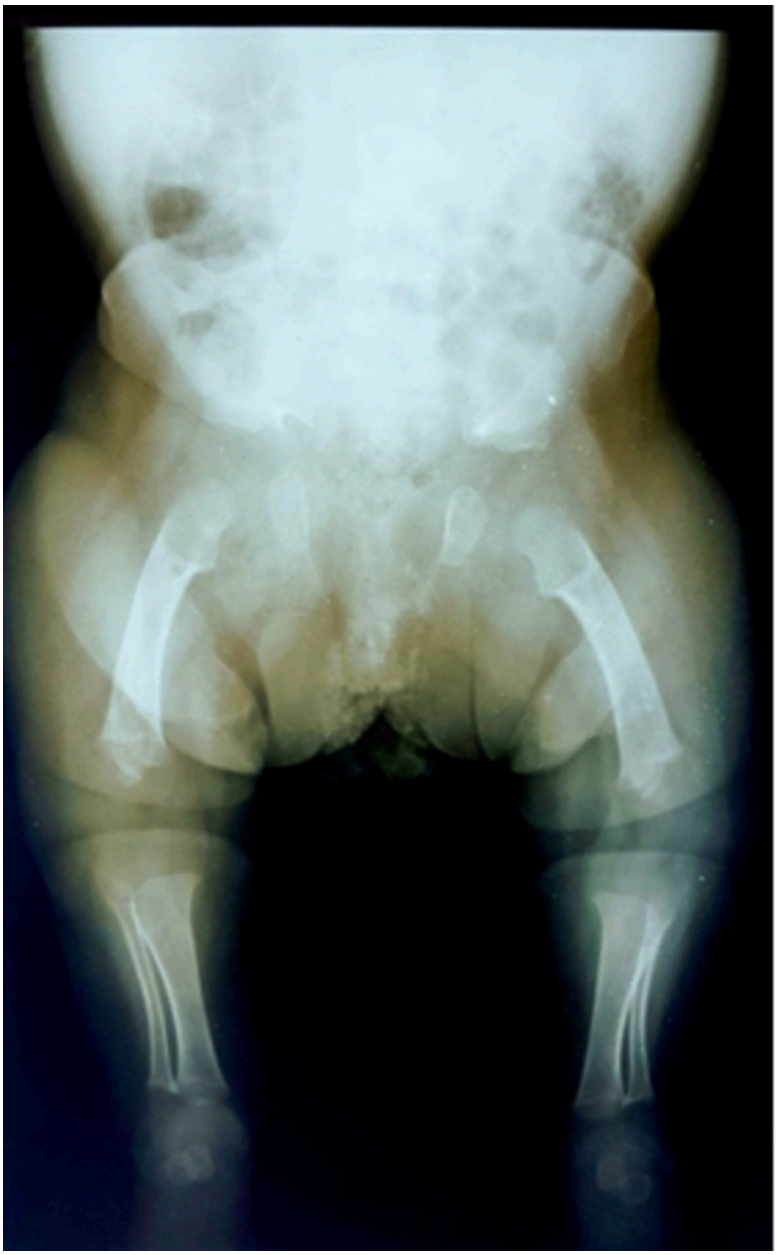

Fig. 2: 02 months year old girl presented with growth disorder. Plain radiography shows rhizomelic shortening of the femur with metaphyseal splaying.

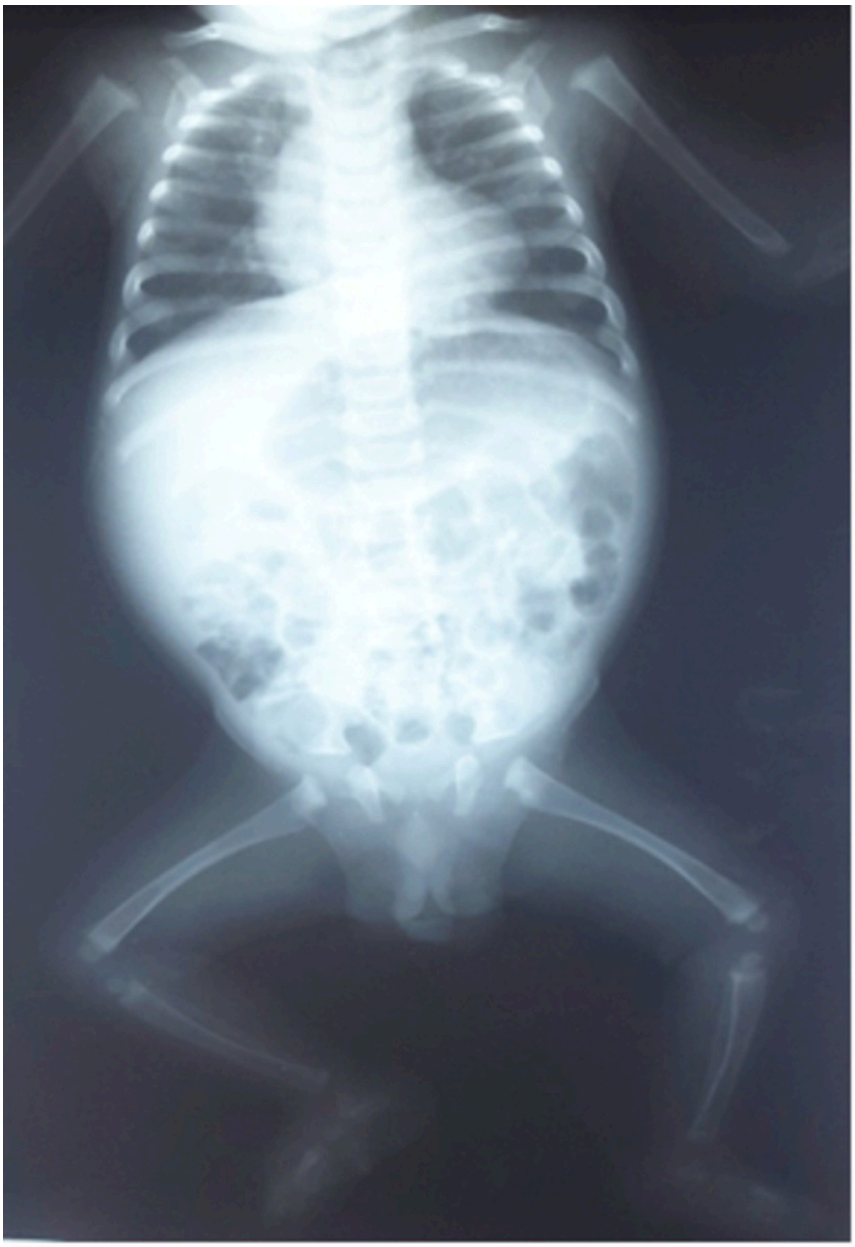

Fig. 3: 06 months year old boy, presented with short stature. Plain radiography shows rhizomelic shortening of the humerus and femur and shortened ribs.

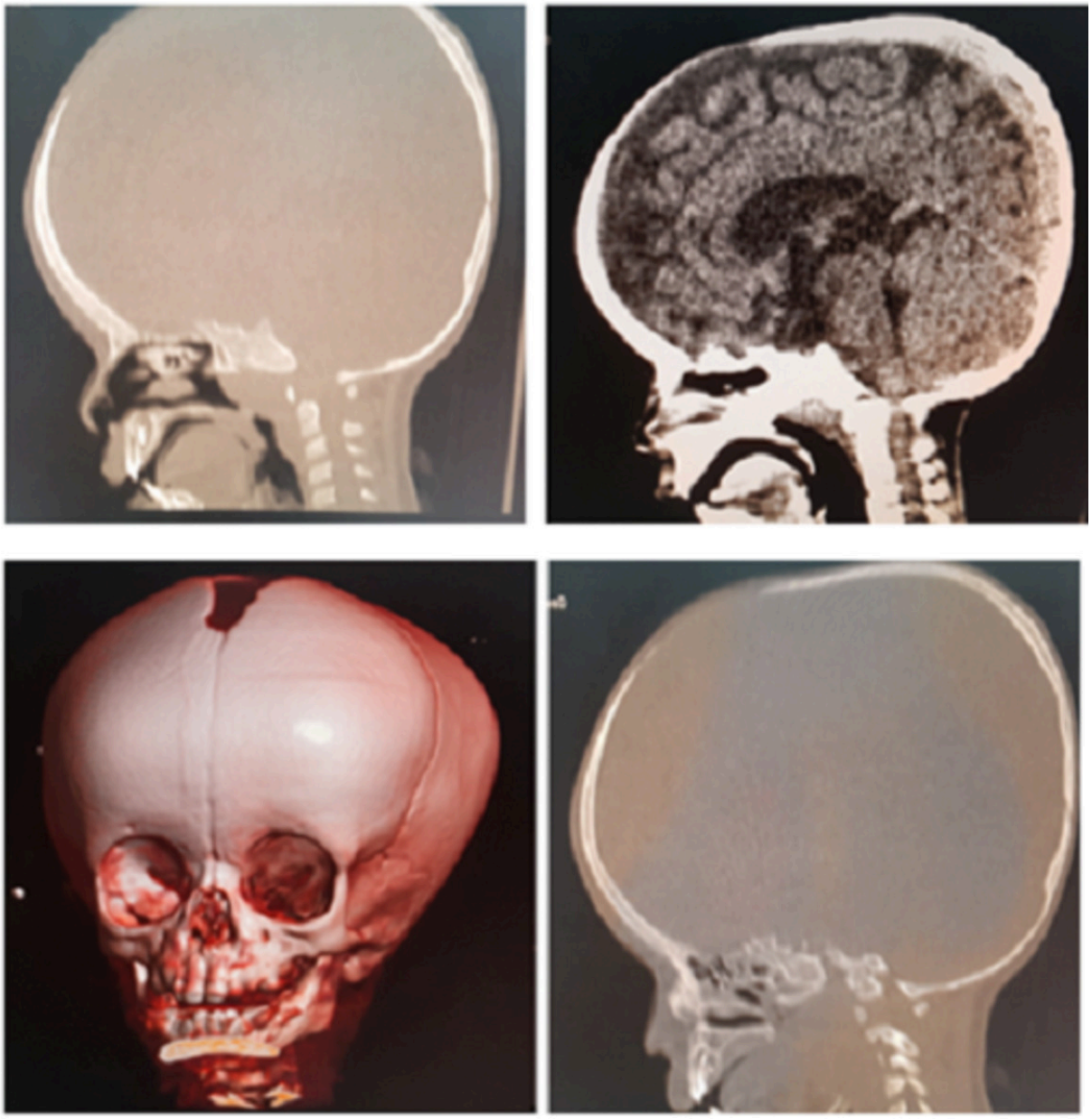

Fig. 4: 10 months old boy with achondroplasia : skull vault enlargement with small skull base and stenosis of the foramen magnum and compression on the cervicomedullary junction.

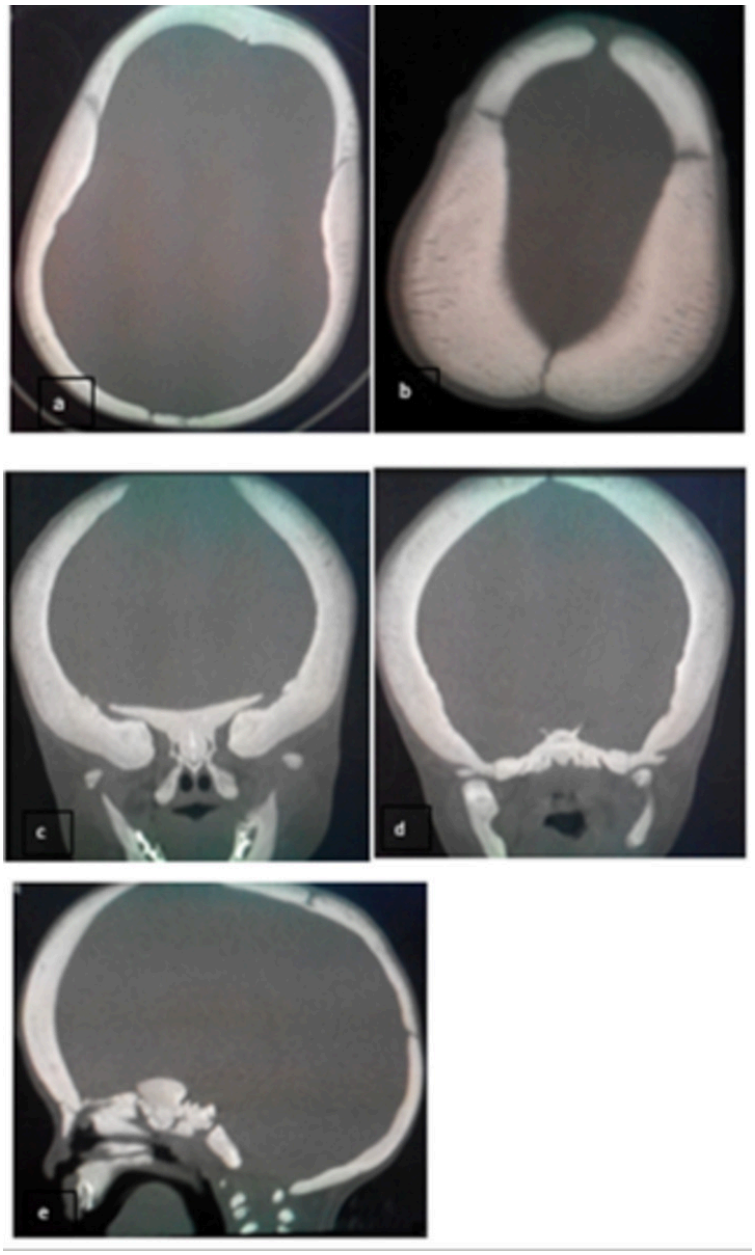

Fig. 7: Head non contrast CT : axial (a,b) , coronal (c,d) and sagittal (e) : osteosclerosis and thickening of the cranial vault

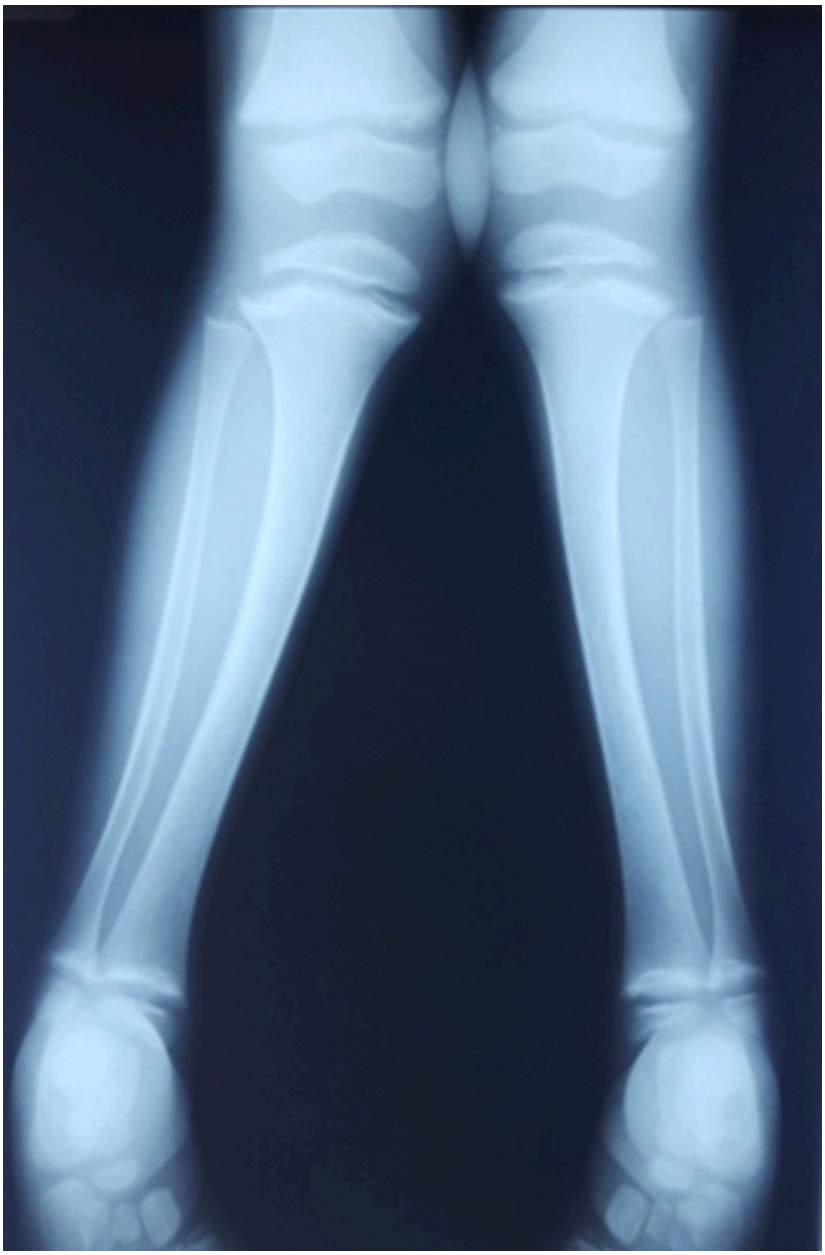

Fig. 5: Metaphyseal chondrodysplasia, Schmid type. Plain radiography in AP projection of both lower limbs. Irregularity and enlargement of metaphysis in knees with genu varum.

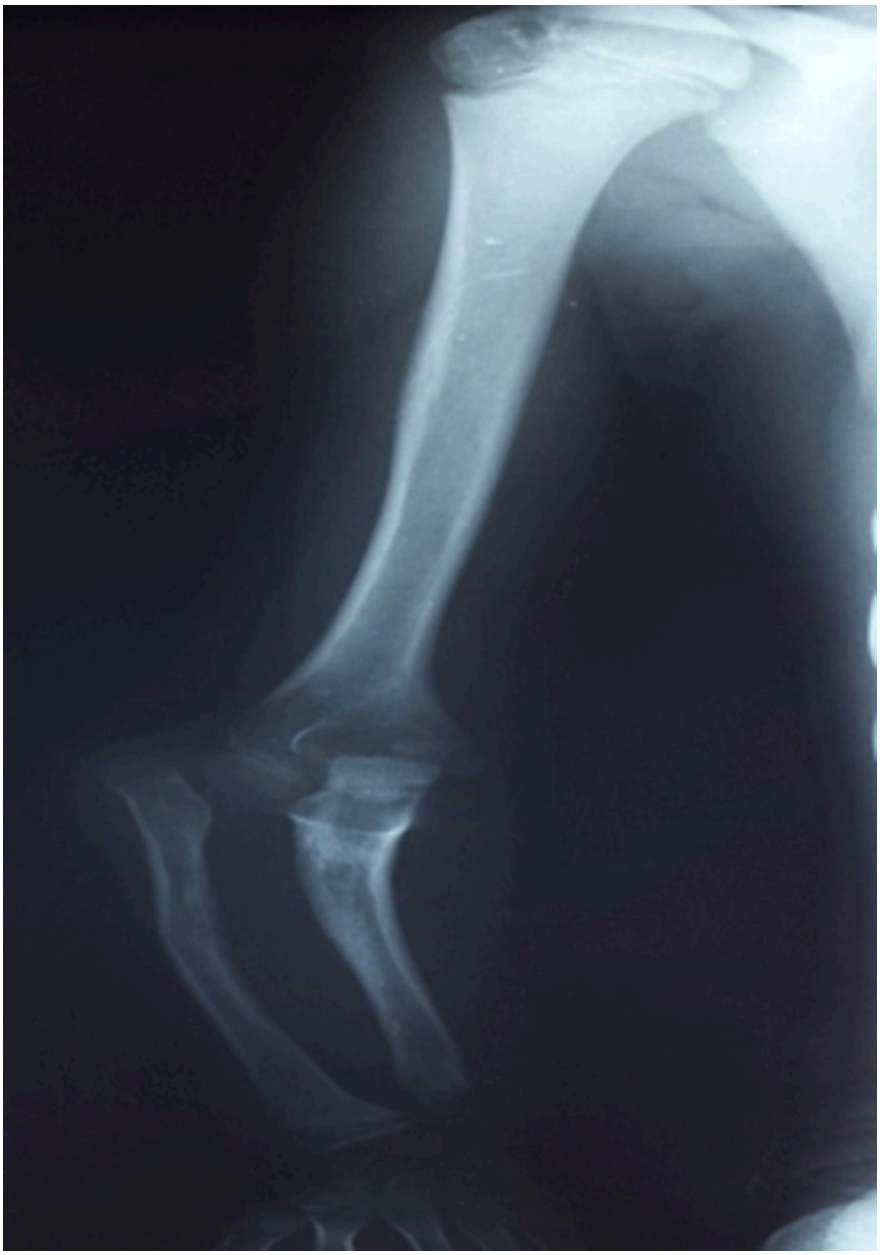

Fig. 6: Madelung deformity: Increased interosseous distance radius to ulna with aberrant curvature of both bones.

Conclusion

Congenital bone disease is a wide spectrum of pathology that may include many associated entities. Plain radiography is the main imaging modality that lead in most cases in presence of clinical symptoms to the right diagnosis.

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