Journal of Cardiovascular Disease

ISSN: 0975-3583, 0976-2833 VOL13, ISSUE 08, 2022

TITLE: SPECTRUM OF VARIOUS TYPES OF SKELETAL DYSPLASIAS IN A TERTIARY CARE HOSPITAL.

[1]Dr. Swapna Ch, [2*]Dr. Vignesh N, [3]Dr Murali S, [4]Dr Sravya Koluguri,

[1]Associate professor, Department of Radiodiagnosis, Kakatiya medical college, Warangal.

[2]Senior Resident, Department of Radiodiagnosis, Kakatiya medical college, Warangal

Junior Resident, Department of Radiodiagnosis, Kakatiya medical college, Warangal.

Junior Resident, Department of Radiodiagnosis, Kakatiya medical college, Warangal.

***Corresponding author**

Dr. Vignesh N,

Senior Resident,

Department of Radiodiagnosis,

Kakatiya medical college, Warangal

ABSTRACT

Introduction: The skeletal dysplasias are a group of more than 450 heritable disorders of bone. They frequently present in the newborn period with disproportion, radiographic abnormalities, and occasionally other organ system abnormalities.

Aims: To enumerate the radiological features of various types skeletal dysplasias.

Materials and methods: It's a Prospective study conducted for a period of one year in patients referred from orthopedic and paediatric departments who were suspected to have skeletal dysplasias. Illustrations of common Skeletal Dysplasias and Dysostoses seen in the study.

Results: A total of 28 cases of skeletal dysplasia and dysostosis were identified in our study. Of these 28 cases 16 were in less than 10 years old, 10 were in the 11-20 years old age group and 2 were in the 21-30 year age group. Of these 28 cases 17 were males and 11 were females. The various types of skeletal dysplasias and dysostoses seen in our study includes Achondroplasia (7%), Chondrodysplasia Punctata(3.5%), Multiple Hereditary Osteochondromas (7%), Enchondroma (3.5%), Fibrous Dysplasias (10.7%), Proximal Focal Femoral Deficiency (3.5%), Isolated Epiphysial Dysplasia of femoral head (3.5%) ,Osteogenesis Imperfecta (7%), Radial Ray Anomaly (10.7%), Radioulnar Synostosis(3.5%), Brachydactyly (7%), Fibular Hemimelia (3.5%), Polydactyly Syndactyly (10.7%), Craniosynostosis, Lunotriquetral Fusion (3.5%), Dysostosis Multiplex (7%).

Conclusion: Precise identification of the tye of skeletal dysplasia is paramount for proper genetic counseling. Postnatal examination and detailed radiographic examination of the fetus especially of the pelvis, limbs, skull and spine are essential to identify the type of skeletal dysplasia.Algorithmic approach of radiologist to either to diagnose a dysplasia or to help the clinician to an appropriate diagnosis.

Keywords: Skeletal dysplasias, Achondroplasia, Chondrodysplasia Punctata, Multiple Hereditary Osteochondromas, Enchondroma.

INTRODUCTION:

Skeletal dysplasias are a heterogeneous group of conditions associated with various abnormalities of the skeleton. These conditions are caused by widespread

ISSN: 0975-3583, 0976-2833 VOL13, ISSUE 08, 2022

disturbance of bone growth, beginning during the early stages of fetal development and evolving throughout life that can cause significant morbidity and mortality [1]. Currently more than 450 different entities have been described based on radiologic, molecular and biochemical criteria [2]. Dysostoses are malformations of single or multiple bones in combination, are due to abnormal blastogenesis in-utero and phenotypically remain static throughout life. Few of the skeletal dysplasias which are lethal in the immediate neonatal period are usually diagnosed in the antenatal ultrasonograms while the non-lethal ones can also be diagnosed in then but most of them present either in the early infantile period or in the childhood with short stature, growth stunting and other physical deformities.

MATERIALS AND METHODS:

It's a Prospective study conducted from September 2020 to June 2021. This study was conducted in Department of Radiodiagnosis, Kakatiya Medical College/ MGM Hospitals, Warangal. The sampling type is consecutive sampling. The patients referred from orthopedic and paediatric departments who were suspected to have skeletal dysplasias are taken up for study. Skeletal survey (fig/ table-1) is done using 1000mA Prognosys Digital Radiography system and a 500 mA Allengers X-Ray unit with a Fujifilm CR system. Informed written consent is taken from the patient.

Inclusion Criteria: Patients suspected to have skeletal dysplasia. Patients willing to give informed written consent.

Exclusion Criteria: Infective and traumatic causes mimicking skeletal dysplasia and Rickets.

Illustrations of common Skeletal Dysplasias and Dysostoses seen in the study are Skull (AP and lateral), Thoracolumbar spine (AP and lateral), Chest (AP), Pelvis (AP), One upper limb (AP), One lower limb (AP) and Left hand (AP)

RESULTS:

A total of 28 cases of skeletal dysplasia and dysostosis were identified in our study. Of these 28 cases 16 were in less than 10 years old, 10 were in the 11-20 years old age group and 2 were in the 21-30 year age group. Of these 28 cases 17 were males and 11 were females.

Table-1: Incidence of various types of skeletal dysplasia/ dysostosis in the study

	Number of cases	Percentages
Age intervals		
< 10 Yrs	16	57
11 - 20 Yrs	10	33
21 - 30 Yrs	2	7
Gender		
Males	17	61
Females	11	39

ISSN: 0975-3583, 0976-2833 VOL13, ISSUE 08, 2022

Types of skeletal dysplasia		
Achondroplasia	2	7
Chondrodysplasia Punctata	1	3.5
Multiple Hereditary Osteochondromas	2	7
Enchondroma	1	3.5
Fibrous Dysplasias	3	10.7
Proximal Focal Femoral Deficiency	1	3.5
Isolated Epiphysial Dysplasia Of Femoral Head	1	3.5
Osteogenesis Imperfecta	2	7
Radial Ray Anomaly	3	10.7
Radioulnar Synostosis	1	3.5
Brachydactyly	2	7
Fibular Hemimelia	1	3.5
Polydactyly Syndactyly	3	10.7
Craniosynostosis	2	7
Lunotriquetral Fusion	1	3.5
Dysostosis Multiplex	2	7

Of these 28 cases, 13 cases were skeletal dysplasias and 15 were dysostosis. The various types of skeletal dysplasias and dysostoses seen in our study includes Achondroplasia (7%), Chondrodysplasia Punctata(3.5%), Multiple Hereditary Osteochondromas (7%), Enchondroma (3.5%), Fibrous Dysplasias (10.7%), Proximal Focal Femoral Deficiency (3.5%), Isolated Epiphysial Dysplasia of femoral head (3.5%) ,Osteogenesis Imperfecta (7%), Radial Ray Anomaly (10.7%), Radioulnar Synostosis(3.5%), Brachydactyly (7%), Fibular Hemimelia (3.5%), Polydactyly Syndactyly (10.7%), Craniosynostosis, Lunotriquetral Fusion (3.5%), Dysostosis Multiplex (7%). The commonest among the skeletal dysplasias in our study are fibrous dysplasias which include both mono-ostotic and polyostotic types. The commonest dysostoses encountered in our study are the radial ray anomalies and polysyndactylies.

ISSN: 0975-3583, 0976-2833 VOL13, ISSUE 08, 2022

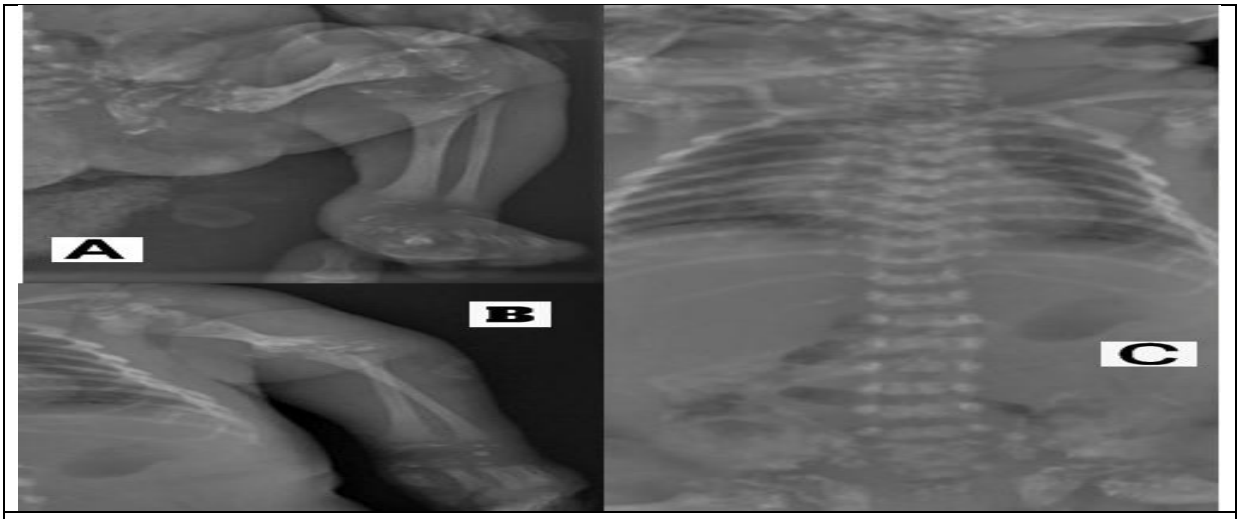

Figure -1: Chondrodysplasia Punctata. (A,B) Upper and lower limb x-rays shows stippling of epiphysis in all large joint. (C) Cropped part of infantogram showing coronal cleft in the vertebral bodies. Note the absence of stippling in the vertebral bodies, hands and feet.

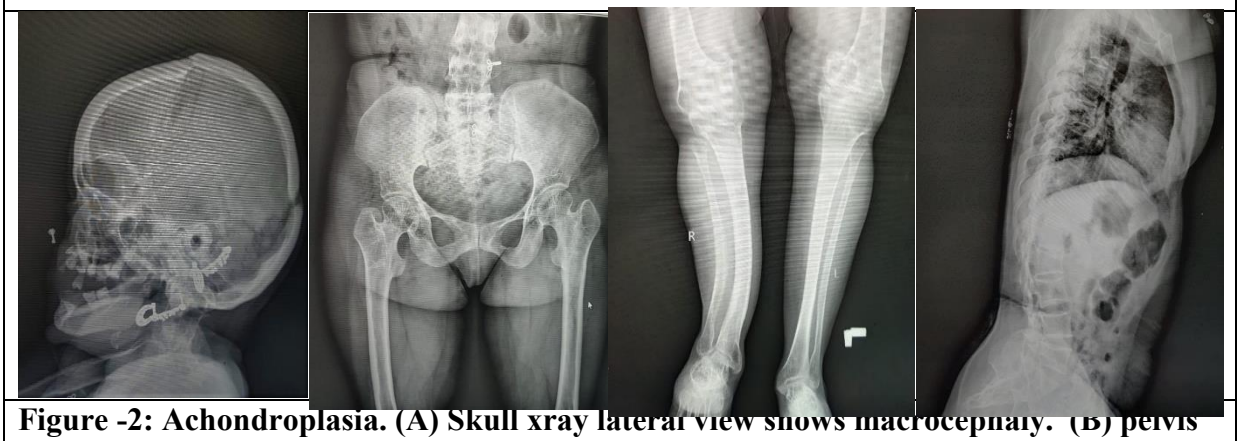

Figure -2: Achondroplasia. (A) Skull xray lateral view shows macrocephaly. (B) pelvis shows champagne glass appearance. (C) Bowing of right tibia and fibula noted. (D) Mild post scalloping noted in the lumbar vertebrae.

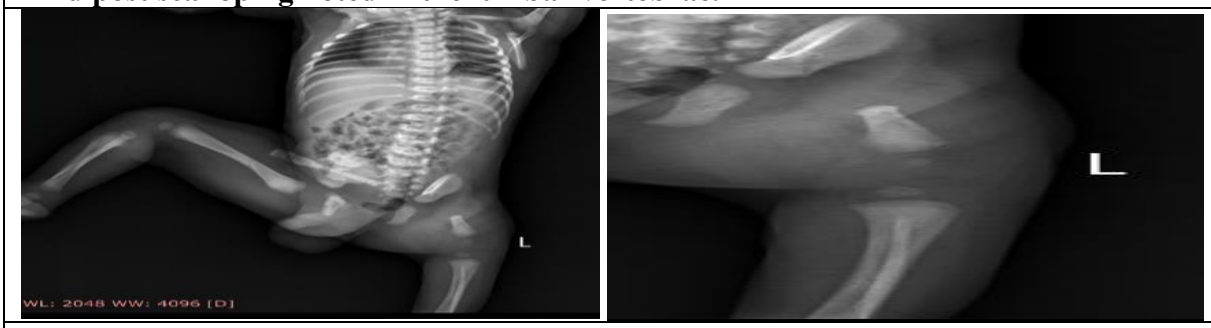

Figure-3: Proximal Focal Femoral Deficiency. Small and dysplastic left femur with absent femoral head and proximal femur with shortening of left lower limb.

ISSN: 0975-3583, 0976-2833 VOL13, ISSUE 08, 2022

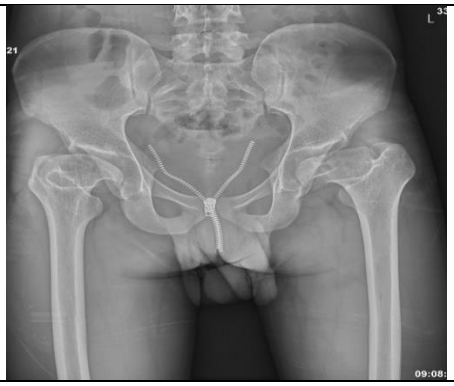

Figure-4: Isolated epiphyseal dysplasia of femur. Bilateral femur shows flattening of epiphysis of head of femur.

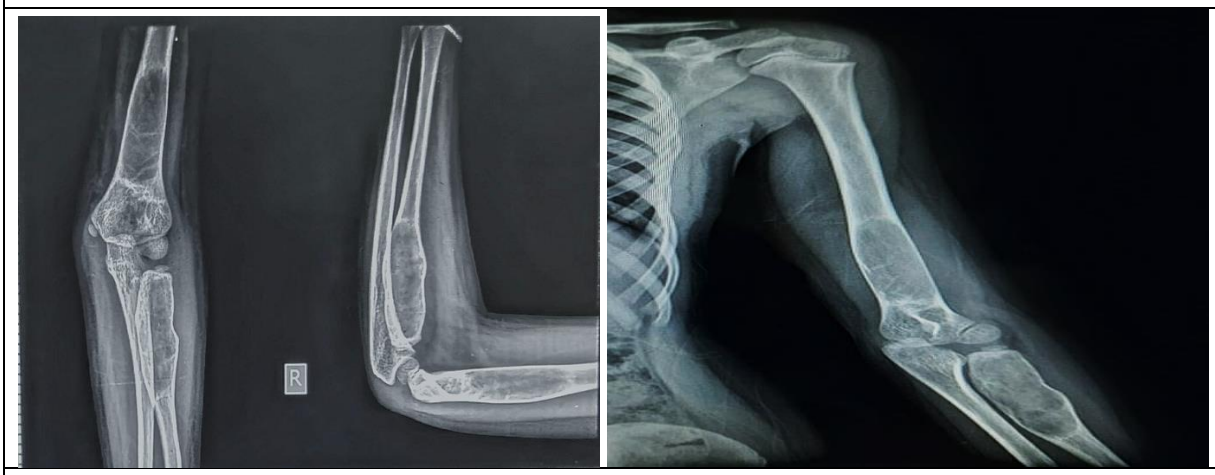

Figure-5: Fibrous Dyaplasia. Metadiaphysial expansile lytic lesion noted involving distal end of left femur and proximal end of left radius with metadiaphysial widending and endosteal scalloping noted

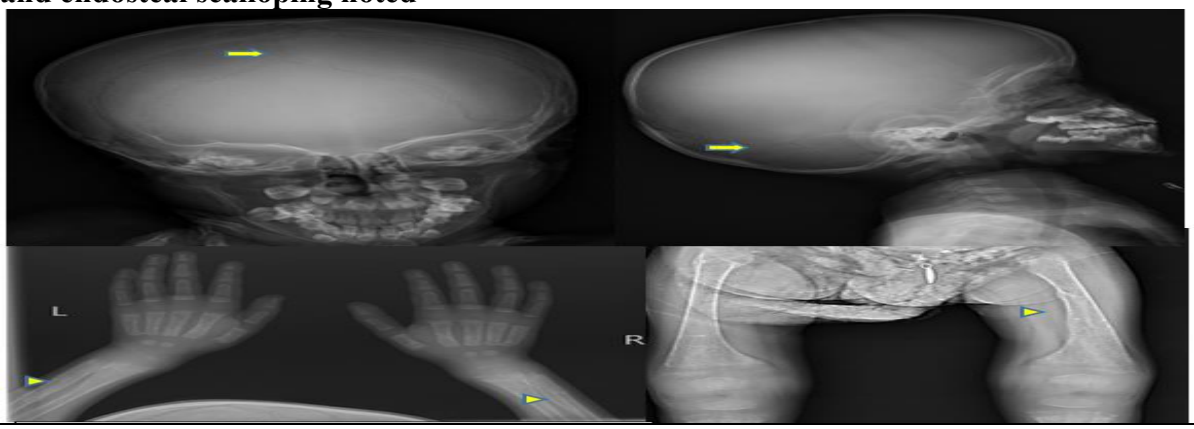

Figure-6 :(A)and (B) skull X ray AP and Lateral show wormian bones. (C) and (D) Multiple fractures noted in long bones Osteopenia, metaphysical flaring

ISSN: 0975-3583, 0976-2833 VOL13, ISSUE 08, 2022

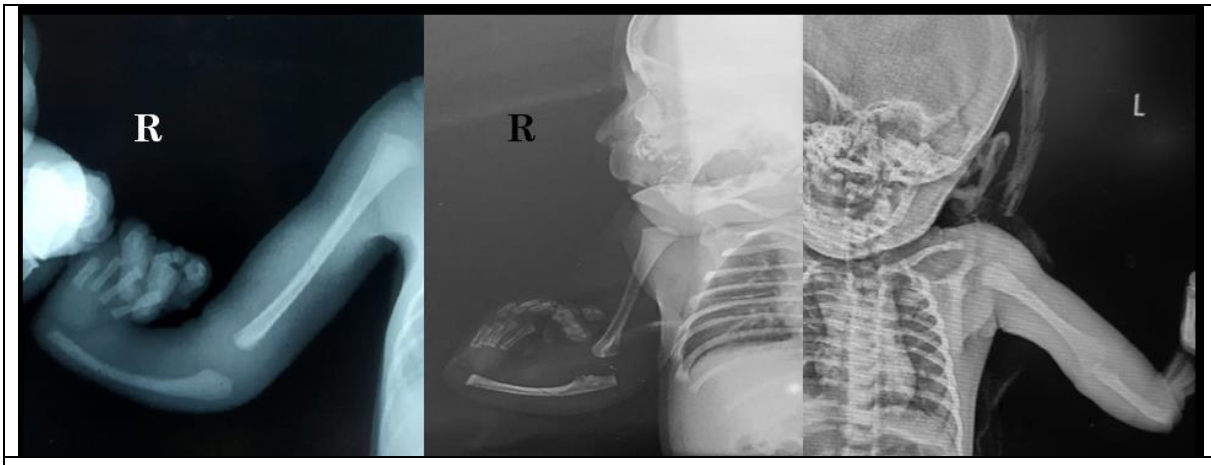

Figure-7: Radial ray anomalies. X ray shows absent radius with absent thumb bones

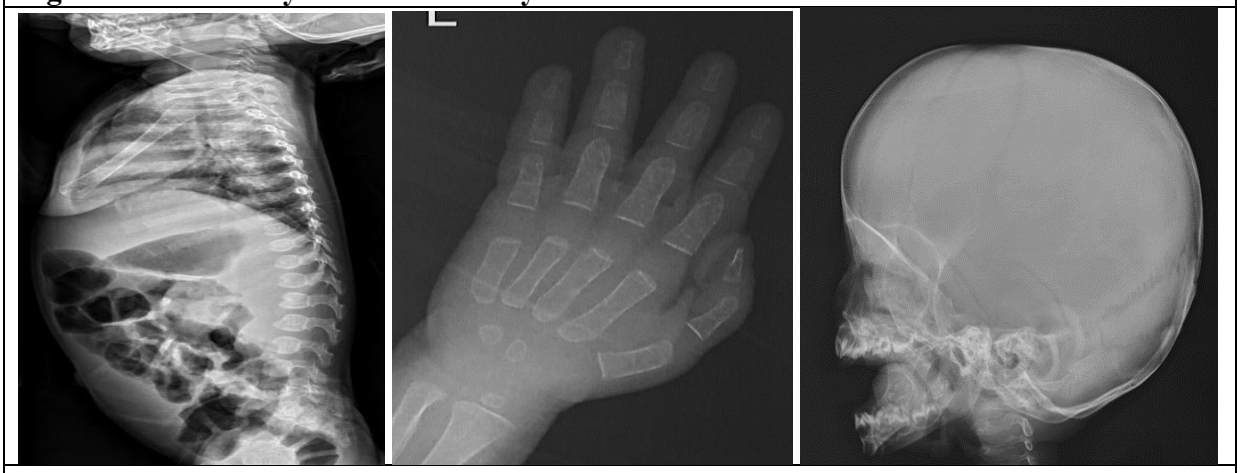

Figure- 8 : Dysostosis Multiplex (A) X-Ray of whole spine shows inferior beaking of vertebrae and platyspondyly of cervical vertebrae (B) Xray hand shows bullet shaped phalanges and proximal pointed metacarpals (C) Xray skull shows 'J' shaped sella.

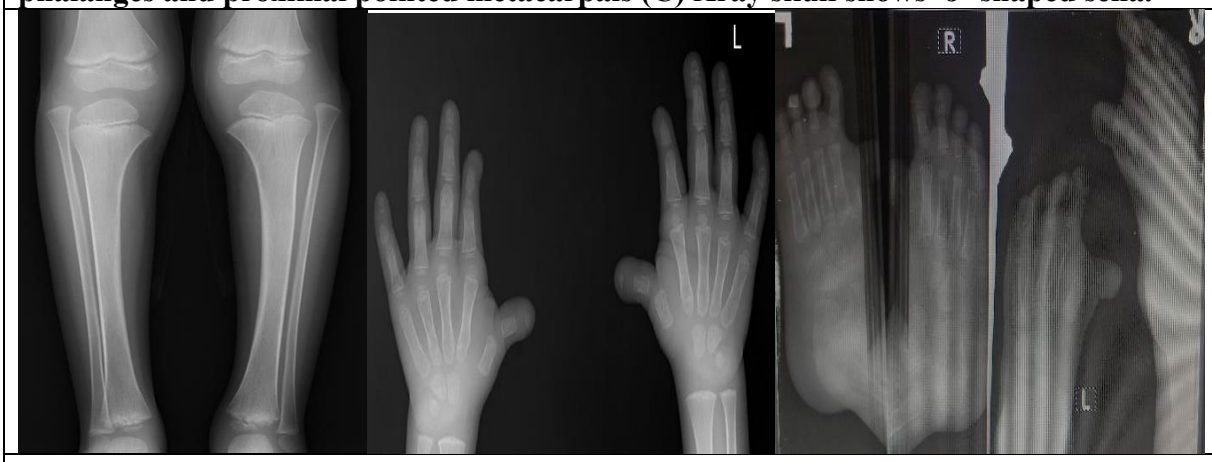

Figure-9 : Brachydactyly – Mononen Type. (A) Mild metaphyseal and epiphyseal irregularity, tibial spurs and relative elongation of the fibulae. (B) and (C) Short first metacarpals and first metatarsals, absent distal phalanges of the index fingers and second toes.

DISCUSSION:

The inheritance of skeletal dysplasias can be autosomal recessive, autosomal dominant, X-linked recessive, X-linked dominant, and Y-linked

ISSN: 0975-3583, 0976-2833 VOL13, ISSUE 08, 2022

manner. Identifying the mode of inheritance is important because it gives information to families regarding future recurrences. Family history, including parental and familial heights and growth patterns, should be obtained from the parents of any affected child to determine if there are similarly affected siblings, or other family members, which can lead to a diagnosis or establish the mode of inheritance. [2]

Children with skeletal disorders are best managed by a multidisciplinary approach that includes obstetricians, pediatricians, neonatologists, medical geneticists, endocrinologists, neurosurgeons, otolaryngologists and orthopaedic surgeons. Sub-speciality care should be directed toward the individual presenting issue. Families should be prepared for ongoing visits to supervise health management.

Chondrodysplasia punctata is of two types Autosomal recessive rhizomelic/lethal variant and X-linked dominant type. The X-linked dominant type (Conradi-Hunermann type) is the common type. Usually congenital CDP is lethal in the first year of birth. The hallmark radiologic feature of CDP is stippling of epiphyses at birth (Figure-1) which later disappers and the epiphysis becomes irregular with asymmetry of bone. CDP can also be seen in warfarin embryotoxicity, in babies born to mothers with auto-immune diseases like systemic lupus erythrematosus [1]. The importance of identifying whether it is rhizomelic/lethal or X-linked dominant type is to determine the prognosis of the patient.

Achondroplasia is the Most common non-lethal skeletal dysplasia. It's a rhizomelic type of dwarfism. It is an Autosomal dominant condition. Antenatally it is difficult to diagnose achondroplastic features until the 3rd trimester. There is shortening of all the long bones and characteristic changes in the pelvic bones, vertebral bodies, hands (Figure-2). There is often a danger of cervical cord compression due to narrowing of the foramen magnum and spinal cord compression. The typical features of achondroplasia are obvious at birth. The most characteristic changes are found in the spine, especially in the lumbar region, pelvis, limbs and skull [3,4].

Proximal focal femoral deficiency is a congenital partial absence of the proximal end of the femur leading to shortening of the entire lower limb (Figure -3). The diagnosis and classification have been based mainly on plain radiograph findings. In severe cases the proximal femur, femoral head and neck, and acetabulum are absent [5]. MR imaging is used to define the cartilaginous proximal femur and the presence or absence of a cartilaginous connection to the femoral head.

Epiphyseal dysplasia is a type of non-rhizomelic dwarfism characterized by flattening and fragmentation of epiphyses (Figure-4). Usually epiphyseal dysplasia involves multiple bones but can be isolated in few cases [5].

ISSN: 0975-3583, 0976-2833 VOL13, ISSUE 08, 2022

Diagnosis is made radiographically with presence of irregular, delayed ossification at multiple epiphyses.

Fibrous dysplasia is a benign lesion in which abnormal fibrous tissue develops in place of normal bone (Figure 5). As it grows in place of normal bone it can lead to fracture or deformity [6]. It can be of two types: Monostotic fibrous dysplasia—only one bone is affected. This is the most common form. Polyostotic fibrous dysplasia—more than one bone is affected[7] .

It is characterized by the development of multiple osteochondromas involving the skeletal system. Usually osteochondromas stop growing with the fusion of growth plate, any growth after that should rise the suspicion of malignancy [8]. other features of malignant transformation include new Lucency, additional scintigraphic activity, cortical destruction, pain after puberty, soft tissue mass, thickened cartilage cap greater than 1.5 cm.

Osteogenesis imperfecta (OI) is an autosomal dominantly or recessively inherited genetic disorder due to mutations in type 1 procollagen genes, characterised by decreased bone mass and increased bone fragility. Severity varies widely from perinatal lethality (type Ⅱ) to milder forms with minimal fractures. Extraskeletal manifestations like blue sclerae, dentinogenesis imperfecta and deafness are also seen [9]. Essential radiological features: (1) Radiologically, OI is characterised by a triad of diffuse osteopenia, pencil-thin cortices, and multiple bony fractures(Figure-6) . The fractures are usually multiple and heal with exuberant callus formation giving rise to "pseudotumour" formation. Associated findings include deformities and pseudoarthrosis; (2) The vertebrae are also osteopenic, have a biconcave "codfish vertebrae" appearance with areas of collapse (3) The skull shows multiple wormian bones, lucent calvarium, enlarged sinuses and platybasia; and (4) The pelvis is also abnormal in shape with deformities like protusio acetabuli and "shepherd crook" femurs [10].

Radial ray anomalies range from radial hypoplasia to radial aplasia with or without accompanying deficiency of the thumb bones(Figure -7). Based on the length of radius it is classified into four types. type I: radius is slightly short and proximal radius usually unaffected and defined as a distal radial physis >2mm proximal to the distal ulnar physis. type II: radius is grossly short and the ulna curves laterally to support the wrist. type III: radius is partially absent. type IV: radius is completely absent [11]. It can be associated with other syndromes like Holt-Oram syndrome, thrombocytopenia absent radius (TAR) syndrome, Fanconi anemia and VACTERL.

Dysostosis Multiplex (Mucopolysaccharidoses) is associated with absence of lysososomal enzymes required for degradation of glycosaminoglycans. There is secondary deposition of GAGs in various tissues causing coarse facies, mental retardation and hepatosplenomegaly [12]. The group of radiographic changes characteristic of MPS (Figure-8) is termed as Dysostosis multiplex. Malformation of the skull, spine, pelvis, chest, long bones, and hands are seen hurler's disease and morquio's disease.

ISSN: 0975-3583, 0976-2833 VOL13, ISSUE 08, 2022

The term brachydactyly indicates shortening of digits due to abnormal development of phalanges, metacarpals, or both [13]. Mononen type of brachydactyly has been reported in literature in 6 patients. The phenotype features include mild short stature, widely spaced nipples, abducted short thumbs, short index fingers, short and abducted great toes and mild bowing of legs (Figure-9). The radiographic features are hypoplastic thumbs and great toes with short first metacarpals and first metatarsals, absent distal phalanges of the index fingers and second toes, mild metaphyseal and epiphyseal irregularity, tibial spurs and relative elongation of the fibulae [14].

Limitations of the study:

This study involves a small sample volume which cannot be generalized to the general population. Hence, the prevalence in the study may not reflect the actual prevalence in the population.

Ethical Committee: Institutional ethical committee clearance obtained for the study.

Conflicts of interest: None

Funding involved in the study: None

CONCLUSION:

Diagnosing of Skeletal dysplasias and dysostoses are not that uncommon in the clinical practice due to the increased availability of radiological facilities. We, the radiologists should have an algorithmic approach to either to diagnose a dysplasia or to help the clinician to an appropriate diagnosis. The correct diagnosis is very much essential for the management of present child and also for counselling parents about future pregnancies and their outcomes. Most skeletal dysplasias and dysostoses can be identified even antenatally by ultrasonography and adequate management can be done.

REFERENCES:

1) Panda A, Gamanagatti S, Jana M, Gupta AK. Skeletal dysplasias: A radiographic approach and review of common non-lethal skeletal dysplasias. 2014 Oct 28
2) Dighe M, Fligner C, Cheng E, Warren B, Dubinsky T. Fetal skeletal dysplasia: an approach to diagnosis with illustrative cases. Radiographics. 2008 Jul-Aug;28(4):1061-77..
3) Pauli, Richard M. "Achondroplasia: a comprehensive clinical review." *Orphanet journal of rare diseases:2019:* vol. 14,1 1. 3.
4) Jana M, Nair N, Gupta AK, Kabra M, Gupta N. Pelvic radiograph in skeletal dysplasias: an approach. Indian J Radiol Imaging. 2017;27:187-99.
5) Berry's Comprehensive Textbook Of Diagnostic Radiology 3rd ed Jaypee Brothers Medical Publishers (P) Ltd, 2021

ISSN: 0975-3583, 0976-2833 VOL13, ISSUE 08, 2022

6) Yochum and Rowe's essentials of skeletal radiology 3rd ed Terry R Yochum; Lindsay J Rowe Lippincott Williams & Wilkins, 2005.

7) Hillock RW, Zupan C: Fibrous dysplasia. Orthopaedic Knowledge Online Journal, 2007; 5(4). February 2016.

8) Grainger & Allison's Diagnostic Radiology 6th Edition Andy Adam, Adrian Dixon, Jonathan Gillard. Elsevier (2016)

9) Renaud, Armelle et al. "Radiographic features of osteogenesis imperfecta." Insights into imaging:2013: vol. 4,4: 417-29.

10) Rauch F, Glorieux FH. Osteogenesis imperfecta. *Lancet* 2004; 363: 1377-1385 [PMID: 15110498 DOI: .1016/S0140-6736(04)16051-0]

11) Sevilla-Montoya R, Aguinaga M, Martínez A, Razo G, Molina B, Frías S, Grether P. Heterogeneous Diagnoses Underlying Radial Ray Anomalies. Indian J Pediatr. 2017 Mar;84(3):200-205.

12) Hendriksz CJ, Harmatz P, Beck M, Jones S, Wood T, Lachman R,. Review of clinical presentation and diagnosis of mucopolysaccharidosis IVA. *Mol Genet Metab* 2013; 110: 54-64

13) Biesecker, Leslie G et al. "Elements of morphology: standard terminology for the hands and feet." American journal of medical genetics. 2009:Part A vol. 149A: 93-127.

14) Tarja K. Mononen, Pamela S. Karnes, Melvin 0. Senac, Jr. , New Skeletal Dysplasia With Unique Brachydactyly. American Journal of Medical Genetics 1992:42:706-713.