

Clinical & Radiological Approach to the Diagnosis of Skeletal Dysplasias

Course Objectives

After attending this course, delegates will:

- 1) have a grounding in the clinical, radiological, genetic and orthopaedic principles underlying skeletal dysplasia diagnosis and management
- 2) be able to diagnose commoner skeletal dysplasias
- 3) have a framework for approaching rarer disorders based on clinical or radiological presentation

Programme

Day 1: Monday 20th May 2024

Refreshments on arrival 9.00-10.00

10.00-10.10	Introduction	CMH
10.10-10.45	Clinical Approach to Skeletal Dysplasias	EK
10.45-11.20	Genetics Overview of Skeletal Dysplasias	SM
11.20-11.55	The Radiology of Skeletal Dysplasias	JB
11.55-12.00	Comfort break with coffee/tea	
12.00-12.30	Prenatal presentation of skeletal dysplasias	JC
12.30-13.00	Orthopaedics for skeletal dysplasias	ML
13.00-13.10	How the course will run	ACO
13.10-14.00	Lunch & Team Photo	
14.00-17.10	Relatively Common Dysplasias with short stature	
14.00-14.10	Case based self-assessment	AS/AC
14.10-14.45	Achondroplasia (<i>FGFR3</i>)	AC/MC
14.45-15.20	Spondyloepiphyseal dysplasia congenita (<i>Col2</i>)	CMH/SM
15.20-15.40	Pseudoachondroplasia (<i>COMP</i>)	JB/EK
15.40-16.00	Coffee/Tea	
16.00-16.25	Cartilage hair hypoplasia (MCD type McKusick)	CMH/SM
16.25-16.40	Diastrophic dysplasia	ACO/JC
16.40-17.00	Asphyxiating thoracic dysplasia/ chondroectodermal dysplasia	AC/EK
17.00-17.10	Dyschondrosteosis (Leri-Weil)	JB/JC
17.10-17.40	Case based review Day 1	AS/AC



Day 2: Tuesday 21st May 2024

<u>Clinical Presentations</u>		
08.45-08.55		Case based self-assessment <i>JB/MC</i>
08.55-09.30	1	Lethal skeletal dysplasias <i>CMH</i>
	2.	The child presenting with fractures
09.30-10.25		Reduced bone density <i>ACO/MC</i>
10.25-11.10		Increased bone density <i>CMH/SM</i>
11.10-11.30		<i>Coffee/Tea</i>
	3.	The child presenting with pain/limp
11.30-12.00		Predominantly epiphyseal dysplasias <i>ACO/AS</i>
12.00-12.30		Spondyloepiphyseal dysplasias <i>JB/JC</i>
12.30-13.00		Case based review <i>JB/MC</i>
13.00-13.50		<i>Lunch</i>
13.50-14.00		Case based self-assessment <i>JC/ACO</i>
	4.	The child presenting with coarse facies/regression
14.00-14.30		Storage disorders <i>JB/EK</i>
14.30-15.15	5.	The child presenting with lumps/bumps/ asymmetry <i>AC/AS</i>
15.15-15.45		Case based review <i>JC/ACO</i>
15.45-16.50		Radiological & Clinical Signs Quiz with <i>coffee/tea</i> <i>CMH</i>
16.50-17.30		Unknown/interesting cases

Course Dinner: 7.30pm – Venue TBC



@SDG4TR

Day 3: Wednesday 22nd May 2024

08.50-09.00		Case based self-assessment	<i>CMH/EK</i>
09.00-09.30	6.	The child with bowed legs	<i>ACO/MC</i>
09.30-10.00	7.	The child with short digits	<i>CMH/SM</i>
10.00-10.30		Case based review	<i>CMH/EK</i>

Radiological Presentations

10.30-10.40		Case based self-assessment	<i>SM/AC</i>
10.40-11.15	1.	Stippling	<i>AS</i>
11.15-11.30		<i>Coffee/Tea</i>	
11.30-12.00	2.	Dumbbell long bones	<i>ACO/JC</i>
12.00-12.30	3.	Under and over modelling	<i>JB/MC</i>
12.30-13.15		<i>Lunch</i>	
13.15-14.00	4.	Dislocations	<i>ACO/AS</i>
14.00-14.30	5.	Craniosynostosis	<i>AC/EK</i>
14.30-15.00		Case based review	<i>SM/AC</i>
15.00-15.15		<i>Coffee/Tea</i>	
15.15-16.45		Final Quiz/Presentation of Prizes	<i>AC/CMH/ACO</i>

End of meeting

This course has achieved 19 CPD credits in accordance with the CPD scheme of the Royal College of Radiologists

FACULTY:

Dr Joy Barber	JB	Radiology
Dr Alistair Calder	AC	Radiology
Dr Jennifer Campbell	JC	Clinical Genetics
Dr Moira Cheung	MC	Paediatric endocrinology
Prof Christine Hall	CMH	Radiology
Dr Esther Kinning	EK	Clinical Genetics
Dr Mark Latimer	ML	Orthopaedics (appendicular)
Prof Sahar Mansour	SM	Clinical Genetics
Prof Amaka C Offiah	ACO	Radiology
Dr Ataf Sabir	AC	Clinical Genetics

