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 11th May 2020

Refreshments on arrival 9.00-10.00
Please submit your unknown/in cases to ACO during the breaks

<table>
<thead>
<tr>
<th>Time</th>
<th>Session</th>
<th>Speaker(s)</th>
</tr>
</thead>
<tbody>
<tr>
<td>10.00-10.10</td>
<td>Introduction</td>
<td>CMH</td>
</tr>
<tr>
<td>10.10-10.50</td>
<td>Clinical Approach to Skeletal Dysplasias</td>
<td>EK</td>
</tr>
<tr>
<td>10.50-11.30</td>
<td>Genetics Overview of Skeletal Dysplasias</td>
<td>SM</td>
</tr>
<tr>
<td>11.30-12.10</td>
<td>The Radiology of Skeletal Dysplasias</td>
<td>JB</td>
</tr>
<tr>
<td>12.10-12.50</td>
<td>Orthopaedics for Skeletal Dysplasias</td>
<td>AS</td>
</tr>
<tr>
<td>12.50-1.10</td>
<td>How the course will run</td>
<td>ACO</td>
</tr>
<tr>
<td>1.10-2.10</td>
<td>Lunch</td>
<td></td>
</tr>
<tr>
<td>2.10 – 5.10pm</td>
<td>Relatively Common Dysplasias with short stature</td>
<td></td>
</tr>
<tr>
<td>2.10-2.45</td>
<td>Achondroplasia (FGFR3)</td>
<td>AC/MC</td>
</tr>
<tr>
<td>2.45-3.20</td>
<td>Spondyloepiphyseal dysplasia congenita (Col2)</td>
<td>CMH/SM</td>
</tr>
<tr>
<td>3.20-3.40</td>
<td>Pseudoachondroplasia (COMP)</td>
<td>GN/JC</td>
</tr>
<tr>
<td>3.40-4.05</td>
<td>Tea</td>
<td></td>
</tr>
<tr>
<td>4.05-4.25</td>
<td>Cartilage hair hypoplasia (MCD type McKusick)</td>
<td>CMH/SM</td>
</tr>
<tr>
<td>4.25-4.40</td>
<td>Diastrophic dysplasia</td>
<td>ACO/JC</td>
</tr>
<tr>
<td>4.40-5.00</td>
<td>Asphyxiating thoracic dysplasia/</td>
<td>CMH/SM</td>
</tr>
<tr>
<td></td>
<td>chondroectodermal dysplasia</td>
<td></td>
</tr>
<tr>
<td>5.00-5.10</td>
<td>Dyschondrosteosis (Leri-Weil)</td>
<td>JB/MC</td>
</tr>
<tr>
<td>5.10-6.00</td>
<td>Case-Based Review Day 1</td>
<td>ALL FACULTY</td>
</tr>
</tbody>
</table>
2: Tuesday 12th May 2020

8.45-9.45  Presentation of unknown/interesting cases  ALL FACULTY

9.45-10.15  Prenatal presentation of skeletal dysplasias  SM

10.15-10.45  Lethal skeletal dysplasias  CMH

10.45-11.05  Coffee

Clinical Presentations

11.05-12.15  1. The child presenting with fractures

11.05-12.00  Reduced bone density  ACO/MC
12.00-12.45  Increased bone density  GN/SM

12.45-1.45  Lunch

1.45-3.00  2. The child presenting with pain/limp
1.45-2.05  Predominantly epiphysseal dysplasias  AC/EK
2.05-3.00  Spondyloepiphysseal dysplasias  JB/JC

3.00-3.30  3. The child presenting with coarse facies/regression

3.00-3.30  Storage disorders  CMH/EK

3.30-3.45  Tea

3.45-4.30  4. The child presenting with lumps/bumps/asymmetry  ACO/JC

4.30-5.30  Case-based review of Day 2  ALL FACULTY

Course Dinner: 7.30pm
Otto’s Restaurant
344 Sharrow Vale Road, Sheffield, S11 8ZP
Day 3: Wednesday 13th May 2020

8.45-9.45  Clinical Presentations (contd)

8.45-9.15  5. The child with bowed legs  ACO/MC

9.45-10.00  Coffee

10.00-11.00  Radiological Signs Quiz  CMH

Radiological Presentations

11.00-11.30  1. Stippling  GN/SM
11.30-11.55  2. Dumbbell long bones  JB/JC
11.55-12.25  3. Undermodelling  CMH/EK
12.25-1.25  Lunch

1.25-2.00  4. Dislocations  ACO/JC
2.00-2.30  5. Craniosynostosis  AC/MC
2.30-3.30  Revision of Day 3/Tea  ALL FACULTY
3.30-5.00  Final Quiz/Presentation of Prizes  ACO

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
Prof Christine Hall  CMH  Radiology
Dr Esther Kinning  EK  Clinical Genetics
Prof Sahar Mansour  SM  Clinical Genetics
Prof Gen Nishimura  GN  Radiology
Dr Amaka C Offiah  ACO  Radiology
Mr Anish Sanghrajka  AS  Orthopaedic Surgery