

# IOF Course - Genetic disorders of bone and their adult expression

## Course Agenda

Day 1	Thursday November 10, 2016	Speaker
09.00-09.05	<b>Welcome</b>	M L Brandi
09:05-09.50	<b>Development, growth and homeostasis of the skeleton</b> <ul style="list-style-type: none"> <li>▪ Skeletal morphogenesis</li> <li>▪ Development and growth of skeletal elements</li> <li>▪ Bone maturation (bone age)</li> <li>▪ The ageing skeleton</li> </ul>	A Superti-Furga A Superti-Furga A Superti-Furga S Ferrari
9.50-10.35	<b>The molecular pathology of genetic skeletal disorders: what do you need to make the skeleton, and what can go wrong?</b> <ul style="list-style-type: none"> <li>▪ Classes of molecules - mineral, structural proteins, enzymes, transporters, signaling factors, vitamins, hormones</li> <li>▪ The many different phenotypes that can be produced by mutations in one single gene - from fetal disease to adult-onset disorders (with examples: hypophosphatasia, OI, chondrodysplasias, osteopetroses, etc)</li> </ul>	L Bonafé  L Bonafé
10.35-10.45	<b>Coffee Break</b>	
From-to	<b>The genetics behind skeletal disease - from rare to common</b> <ul style="list-style-type: none"> <li>▪ Type of mutations</li> <li>▪ The importance of pedigree analysis in making a diagnosis</li> <li>▪ Mechanisms: loss of function, dominant negative, protein suicide, mosaicism</li> <li>▪ How to find the gene: panels, WES, WGS</li> </ul>	S Unger S Unger S Unger S Unger
11.35-12.30	<b>How to obtain and integrate the diagnostic information</b> <ul style="list-style-type: none"> <li>▪ Ask the right questions: age of onset, clinical history, the growth curve</li> <li>▪ Physical signs - not only bone in syndromic disorders</li> <li>▪ Which instrumental diagnostic tools should be used, and how to read them - an art and its pitfalls</li> <li>▪ Don't feel alone and don't keep your cases in a drawer - networking and the internet as invaluable tools for diagnosis</li> </ul>	A Superti-Furga  A Superti-Furga A Superti-Furga  A Superti-Furga

<b>Day 1</b>	<b>Thursday November 10, 2016</b>	<b>Speaker</b>
12.30-13.30	Lunch	
13.30-14.10	<b>Diagnostic clinical approach to bone fragility</b> <ul style="list-style-type: none"> <li>▪ Osteoporosis in the young</li> <li>▪ Differential diagnosis with secondary causes and rare metabolic diseases</li> </ul>	S Ferrari M Javaid
14.10-15.10	<b>Focus on recurrent fracture or avascular necrosis as the presenting sign</b> <ul style="list-style-type: none"> <li>▪ Osteogenesis Imperfecta (OI)</li> <li>▪ Gaucher's</li> <li>▪ Hypophosphatasia (HPP)</li> </ul>	M Javaid A Trombetti M L Brandi
15.10-15.30	Coffee Break	
15.30 - 16.30	<b>Focus on chronic bone pain: could it be a genetic disorder?</b> <ul style="list-style-type: none"> <li>▪ Fibrous dysplasia</li> <li>▪ Melorrrheostosis</li> <li>▪ Camurati-Engelmann</li> <li>-</li> </ul>	M Javaid M Javaid L Masi
16.30 - 17.30	<b>Too much bone: bad or good?</b> <ul style="list-style-type: none"> <li>▪ Osteopetrosis</li> <li>▪ Sclerosteosis/van Buchem disease</li> <li>▪ High bone mass / LRP5 mutations</li> </ul>	S Papapoulos
<b>Day 2</b>	<b>Friday November 11, 2016</b>	
08.30-10.00	<b>Focus on mineral metabolism and its disorders</b> <ul style="list-style-type: none"> <li>▪ Hypoparathyroidism</li> <li>▪ Rare forms of rickets/ osteomalacia</li> </ul> <p>a) Rickets in vitamin D receptor mutation b) Oncogenic Osteomalacia c) XLH</p>	M L Brandi  M Javaid M L Brandi L Masi
10.00-10.15	Coffee Break	
10.20-11.15	<b>Targeted therapies in genetic skeletal disorders</b> <ul style="list-style-type: none"> <li>▪ Mechanisms of action of antiresorptive and anabolic drugs for bone</li> <li>▪ Other “molecular” therapies</li> </ul>	S Ferrari  M L Brandi
11.15-12.00	<b>Review of general diagnostic strategies for adults with rare skeletal disorders</b>	S Papapoulos
12.00-13.30	Lunch	
13.30-16.00	<b>Difficult Cases</b>	All
16.00	<b>Wrap-up comments and course evaluation</b>	M L Brandi