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Skeletal dysplasias are conditions in which there is an abnormality of bone and/or cartilage growth, which may show phenotypic evolution throughout life. They are genetically and radiologically heterogeneous, with accurate diagnosis requiring careful correlation of clinical, genetic and radiological information.
Presentation may be with unexplained short stature, and this, in the presence of disproportion is the most common indication for a skeletal survey within paediatric endocrinology. However, it is important that imaging is used with care and that only those patients who need it are selected for skeletal survey.
This session will demonstrate the utility of radiological assessment, and skeletal surveys in particular, in guiding laboratory and genetic tests and in differentiating causes of short stature in children. Important differential diagnoses will be discussed and key results of a systematic review of the applicability of the Greulich and Pyle bone age standard to children of different ethnicities will be highlighted. Given its increasing use, the session will also review the automated bone age software tool, BoneXpert, which not only assesses bone age according to both Greulich and Pyle and Tanner and Whitehouse standards, but also provides an assessment of bone health – the “bone health index”. By the end of the session, delegates should have an understanding of when to investigate for skeletal dysplasia, which images to request and be able to recognise salient features of some relatively common genetic conditions presenting with short stature to the paediatric endocrinologist.