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New taxonomy classifies rare genetic bone disorders by metabolic pathogenesis

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An International Osteoporosis Foundation (IOF) Working Group on Skeletal Rare Diseases has published a new classification of rare genetic metabolic bone disorders (RGMBDs) according to their metabolic pathogenesis.

Skeletal Rare Diseases such as osteogenesis imperfecta, juvenile Paget's disease or osteopetrosis, are only three of the more than 400 developmental abnormalities of the skeletal system that affect patients worldwide. Although rare, these 'orphan' diseases have a very serious, and often devastating, impact on the lives of the individuals affected.

Prof. Maria Luisa Brandi MD, Professor of Endocrinology and Metabolic Diseases at the University of Florence, Italy, lead author and Chair of the IOF Working Group on Skeletal Rare Diseases stated, "Due to the rarity of these diseases diagnosis is a challenge, and most patients - who are often children - currently have few choices when it comes to therapy. To date, the diagnosis of rare skeletal diseases is based primarily on clinical phenotype and radiographic analysis. We believe that knowledge of the metabolic pathway that characterizes these diseases provides important information that will help doctors select the most appropriate pharmacological treatment in patients affected by these complex diseases."

The taxonomy comprises 116 OMIM phenotypes, with 86 affected genes related to bone and mineral homeostasis. It divides the diseases into four major groups by listing the disorders due to:

- altered osteoclast, osteoblast or osteocyte activity;
- altered bone matrix proteins;
- altered bone micro-environmental regulators;
- deranged calciotropic hormonal activity.

The classification is a basis for the creation of an international registry of rare skeletal diseases. It also provides valuable information that may assist researchers in the development of genetic tests based on next generation sequencing, and in the initiation of large intervention trials to assess the efficacy of orphan drugs.

Prof. Cyrus Cooper, Chair of the IOF Committee of Scientific Advisors added, "Genetic disorders of the skeletal system represent a significant number of the world's recognized rare diseases. For affected individuals, these complex conditions are most often severe, degenerative, and chronically debilitating. Unfortunately, due to the rarity of these disorders, patients are currently faced with limited therapeutic options. We hope that knowledge derived from this new taxonomy will comprise an important first step in facilitating international cooperation of research, ultimately leading to new avenues for diagnosis and therapeutic intervention."

More information: Taxonomy of rare genetic metabolic bone disorders, L. Masi, D. Agnusdei, J. Bilezikian, D. Chappard, R. Chapurlat, L. Cianferotti, J.-P. Devolgelaer, A. El Maghraoui, S. Ferrari, K. Javaid, J.-M. Kaufman, U. A. Liberman, G. Lyritis, P. Miller, N. Napoli, E. Roldan, S. Papapoulos, N. B. Watts, M. L. Brandi. *Osteoporos Int*, DOI: 10.1007/s00198-015-3188-9

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