New taxonomy of non-skeletal rare disorders with impact on bone

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Thanks to major progress in the understanding and management of rare congenital diseases and syndromes, many patients with these rare disorders are now living longer lives. With this progress it has become apparent that many non-skeletal rare diseases have an impact on bone mass, bone quality and/or bone metabolism, with potentially severe repercussions for quality of life in adults.

The new paper "Bone fragility in patients affected by congenital diseases non skeletal in origin," published in *Orphanet Journal of Rare Diseases* by the International Osteoporosis Foundation (IOF) Skeletal Rare Diseases Working Group (SRDWG), provides a first taxonomic classification of selected non-skeletal rare congenital disorders with an impact on bone physiology on the basis of phenotypes. The diseases have been described according to the systemic disease; genetic defect; pathophysiology of bone phenotype; and therapy, where available.

The classifications are provided in separate tables for these rare disorders as follows:

- Metabolic rare diseases: lysosomal storage diseases, disorders of sulfur metabolism, disorders of tyrosine pathway
- Liver rare diseases: disorders of copper pathway (Menkes and Wilson disease)
- Respiratory rare disease: cystic fibrosis
- Hematological rare diseases: mastocytosis, beta-thalassemia, hemophilia, sickle cell disease, Ghosal hemato-diaphyseal dysplasia, severe congenital neutropenia, Histiocytosis
- Neurological Rare Disease: Rett Syndrome
- Malformation: Tricho-rhino-phalangeal syndrome, type I

Professor Maria-Luisa Brandi, chair of the past IOF SRD Working Group and convener of the IOF Skeletal Rare Disease Academy, stated:

"With this work, we have established the first classification of systemic rare diseases which alter bone metabolism and which may be amenable to different therapeutic approaches to maintain bone health. We look forward to updating the classification as new knowledge and new therapeutic options become available."

Professor Nicholas Harvey, chair of the IOF Committee of Scientific Advisors (CSA) and the IOF Skeletal Rare Disease Academy, added:

"This new publication, documenting a novel approach to the classification of these rare diseases, demonstrates the enormous value in bringing together global leaders in this field through the International Osteoporosis Foundation's science program, continuing through the IOF Skeletal Rare Diseases Academy, to generate state-of-the-art reports that will inform clinical practice worldwide."

More information: et al, Bone fragility in patients affected by congenital diseases non skeletal in origin, *Orphanet Journal of Rare Diseases* (2021).

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