

Erratum to: Taxonomy of rare genetic metabolic bone disorders

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Correction of publisher's error: In the original publication, Fig. 1 appeared in the place of Fig. 8 and vice versa. The article has now been updated to correct this error.

USA-based reference websites for rare diseases

- National Center for Biotechnology Information, Online Mendelian Inheritance in Men and GeneReviews databases: <http://www.ncbi.nlm.nih.gov>, <http://www.ncbi.nlm.nih.gov/omim> and <http://www.ncbi.nlm.nih.gov/books/NBK1116/>
- Genetic and Rare Diseases Information Center (GARD) by National Institutes of Health: <http://rarediseases.info.nih.gov/gard/browse-by-first-letter/a>
- Rare Disease Database by the National Organization for Rare Disorders (NORD), <https://www.rarediseases.org/rare-disease-information/rare-diseases>
- OMIM, www.omim.org

Europe-based reference websites for rare diseases

- Orphanet, European database dedicated to information on rare diseases and orphan drugs: www.orpha.net
- Swedish rare disease database: <http://www.socialstyrelsen.se/rarediseases>

Fig. 1 Main English websites on rare diseases with constantly updated databases (as most recently accessed in January 2015)

The online version of the updated original article can be found at <http://dx.doi.org/10.1007/s00198-015-3188-9>.

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Biochemical Markers

- Serum Calcium
- Urinary Calcium
- Serum Phosphate
- Urinary Phosphate
- Serum Magnesium
- Urinary Magnesium
- Parathyroid hormone (PTH)
- Fibroblast growth factor 23 (FGF23)
- 25 hydroxyvitamin D₃ [25(OH)D₃]
- 1,25 dihydroxyvitamin D₃ [1-25(OH)₂D₃]
- Bone Formation Markers:
 - Total Alkaline Phosphatase, Bone Alkaline Phosphatase, Osteocalcin, Procollagen 1 N-terminal Propeptide (P1NP), Procollagen 1 C-terminal Propeptide (P1CP)
- Bone Resorption Markers:
 - Hydroxyproline, Pyridinoline and Deoxypyridinoline, Cross-linked N-telopeptide of type I collagen (NTX), Cross-linked C-telopeptide of type I collagen (CTX)
- Bone Mineralization Markers:
 - Pyridoxal-5'-phosphate
 - Urinary 4-pyridoxic acid
- Bone Microenvironment Products:
 - Sclerostin, RANK-ligand, Osteoprotegerin

Instrumental exams

- DEXA: Lumbar Spine, hip, wrist, total body
- Ultrasound (US): heel, finger
- Peripheral Quantitative Computed Tomography (pQCT): leg, wrist
- Quantitative Computed Tomography (QCT): spine
- X-rays
- Radiographic vertebral morphometry
- DEXA: Vertebral Fracture Assessment (VFA)
- Bone scintigraphy
- Positron Emission Tomography (PET)
- Magnetic Resonance Imaging (MRI)

Bone Biopsy

- Histology
- Histomorphometry

In vitro assays

Fig. 8 Biochemical/instrumental exams and in vitro tests for characterizing metabolic bone diseases. Measurements of biochemical indexes and hormones regulating mineral homeostasis can help in confirming or excluding systemic bone metabolic disorders. The assessment of bone turnover is important in order to plan further therapeutic approaches. Bone quantity and quality appraisal and prevalent vertebral fracture assessment may help to refine the metabolic

framing of the disease, manifesting with an otherwise evident bone phenotype, and is crucial in the follow-up of the treated patient. Bone biopsy is critical in selected cases for the identification and for differential diagnosis. In vitro assays can be useful to identify supposed functional abnormalities of bone cells and/or matrix proteins (e.g., in collagen-related disorders)