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## Degrading mutations

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Jonathan B Weitzman

Email: jonathanweitzman@hotmail.com

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**Inherited osteolysis** (also known as vanishing bone syndrome) is characterized by abnormal destruction and resorption of bone. In the July issue of *Nature Genetics*, Martignetti *et al.* identify mutations in the matrix metalloproteinase 2 gene (*MMP2*) in several Saudi Arabian families with **osteolysis** (*Nature Genetics* 2001, **28**:261-265). They performed a genome-wide search for homozygous-by-descent microsatellite markers, in order to hone in on a region of chromosome 16q12 that includes the **MMP2 gene**. *MMP2* enzymatic activity could not be detected in serum or cultured skin fibroblasts from these patients. Martignetti *et al.* identified two homoallelic *MMP2* mutations: a missense mutation (R101H) within the prodomain and a non-sense mutation (Y244X) that deletes the catalytic sites. The authors suggest that the unexpected role of *MMP2* in osteolysis may involve deregulated extracellular matrix breakdown leading to an imbalance between bone synthesis and resorption.

## References

1. Osteolysis, hereditary multicentric, [<http://www.ncbi.nlm.nih.gov:80/entrez/dispomim.cgi?id=259600>]
2. *Nature Genetics* , [<http://genetics.nature.com>]
3. New form of idiopathic osteolysis: nodulosis, arthropathy and osteolysis (NAO) syndrome.
4. Completion of the primary structure of the human type IV collagenase preproenzyme and assignment of the gene (CLG4) to the q21 region of chromosome 16.