Papers of the Week

Creating a Mouse Model for Hajdu Cheney Syndrome

* See referenced article, *J. Biol. Chem.* 2016, 291, 1538–1551

**Hajdu Cheney Mouse Mutants Exhibit Osteopenia, Increased Osteoclastogenesis, and Bone Resorption**

Notch2 is a critical molecule involved in skeletal development and bone remodeling. Mutations in NOTCH2 are associated with a bone disease called Hajdu Cheney syndrome that is characterized by osteoporosis and bone fractures. In this Paper of the Week, Ernesto Canalis at the University of Connecticut Health Center and colleagues created a mouse model for Hajdu Cheney syndrome to better understand the underlying mechanisms responsible for the disease. They introduced a mutation in Notch2 to mimic the mutation found in humans. Compared to wild-type mice, mutant mice were smaller with shorter femurs and had low bone mass by the age of 1 month. Mutant mice had enhanced bone resorption and an increased number of bone-resorbing cells; this was because the precursor pool of bone-resorbing osteoclasts as well as their differentiation toward mature cells was greater in Hajdu Cheney mutant mice. The authors say, “Although one needs to be cautious with the extrapolation of these results to human disease, an increase in bone resorption could explain the pronounced osteoporosis suffered by subjects with Hajdu Cheney syndrome.”

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Tomography images of bone and femurs of mutant and normal mice.
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