RARE BONE DISEASE HIGHLIGHTS FROM THE ASBMR 2022 ANNUAL MEETING: CLINICIAN PERSPECTIVE

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ASBMR
The American Society for Bone and Mineral Research

Therapeutic targets in rare bone disease are evolving, from treatment of symptoms with small-molecule and antibody drugs, to **treatment of underlying genetic drivers** of disease

INSIGHTS ON FOP

- Painful soft tissue swelling that precedes **irreversible heterotopic ossifications** characterize this severe congenital condition
- FOP is caused in nearly all cases by the same variant in the *ACVR1* gene
- Several therapies in development **reduce BMP signaling**, thereby reducing degree of heterotopic ossification

INSIGHTS ON OI

- By treating the underlying genetic lesion, **cell therapy** may promote healthy osteogenesis in OI patients
- Allogeneic **transplantation** of fetal liver-derived mesenchymal stem cells is tested in children with type III and IV OI
- As OI presents **variable natural history**, clinicians should exercise caution when interpreting data on therapeutic effect in early small-cohort studies

INSIGHTS ON HCH

- A **skeletal dysplasia** that has received comparatively less attention, possibly due to its milder disease presentation
- An experimental **mouse model** for HCH has been developed, in which daily infigratinib, a selective FGFR tyrosine kinase inhibitor, shows positive effects on **limb growth** and **proportion**

BMP: bone morphogenetic protein
FGFR: fibroblast growth factor receptor
FOP: fibrodysplasia ossificans progressiva
HCH: hypochondroplasia
OI: osteogenesis imperfecta

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