FD/MAS ALLIANCE

FD/MAS Patient Priorities

Fibrous dysplasia/McCune-Albright syndrome (FD/MAS) is a rare multisystem disease caused by somatic variants in the gene GNAS. The variant results in constitutive activation of the Gsα cAMP signaling pathway in cells. Skeletal manifestations of FD include bone pain, fractures, deformity, and osteomalacia/rickets.

FD/MAS patients prioritize multi-stakeholder, collaborative, and patient-centric progress in developing FD/MAS treatment. The FD/MAS Alliance is a patient-driven advocacy group that fosters the development of evidence-based treatments. See Video

We believe

- *Projects that feature collaborations across multiple institutions should be encouraged.*
- *Reagents and research tools, including animal models, must be freely accessible and deposited in a public repository without restriction.*

Clinical, translational, and mechanistic research studies to address any unmet needs in the care of FD/MAS patients are of the utmost importance. Research priorities for FD/MAS patients include

1. **Mechanisms and treatment of pain:** Understanding the mechanism and treatment of bone pain in FD/MAS remains a significant unmet need. Fulfilling this unmet need would have significant implications for bone diseases in general.

2. **Effective treatment for bone disease:** The basic pathology of FD is that it is a low-grade neoplasm of skeletal stem cells. The biology of this neoplastic process is poorly understood. Studies of skeletal stem cell biology to understand the neoplastic process have widespread implications for skeletal and neoplasia in general.

3. **Dysregulating pathways in FD/MAS:** Signaling pathways that underlie the pain and neoplasia include Gsα/cAMP, protein kinase A and salt-inducible kinase. Development or testing of molecules that target these pathways would advance the understanding and treatment of FD/MAS and related disorders.

4. **Molecular dysregulation:** Molecules that are dysregulated in FD/MAS include RANKL, IL6, cAMP, and FGF23. Studies to target and understand this dysregulation will advance FD/MAS care.

IMPORTANT RESOURCES FOR RESEARCHERS

- FD/MAS Alliance: a community-led 501c3 nonprofit that serves people affected by FD/MAS through programs research, education, and advocacy. We nurture close collaborative efforts between patients and the Scientific and Medical Advisory Councils, whose volunteer members include many world-class scientists and clinicians with expertise in FD/MAS pathophysiology, metabolic bone disease, and endocrinology. Our mission includes the support of research to develop better evidence-based treatments.
- Screening and Natural History of Patients With Polyostotic Fibrous Dysplasia and the McCune-Albright Syndrome - 300 patients.
- The FD/MAS Patient Registry is an IRB-approved FD/MAS Alliance research project with patient-reported data and over 1000 patients.

To learn more, visit fdmasalliance.org or email Adrienne McBride, Executive Director at amcbride@fibrousdysplasia.org (EIN 02-0715210).