DEPARTMENT OF HEALTH & HUMAN SERVICES

Public Health Service

National Institutes of Health
Bethesda, Maryland 20892

The Honorable John F. Kerry
United States Senate
Washington, DC 20510

Dear Senator Kerry:

Thank you for your thoughtful letter in support of the important research on McCune-Albright Syndrome/Fibrous Dysplasia (MAS/FD) being conducted or funded by the National Institutes of Health (NIH). The rarity of this disease does not make it any less difficult for the thousands of families struggling with MAS/FD, and I want to assure you that we are committed to supporting research into both the causes and potential treatments of this terrible syndrome.

As you said in your letter, much of our understanding of MAS/FD is the result of studies done at many of the Institutes and Centers here at NIH, including the discovery of the genetic basis of the disorder by National Institute of Diabetes and Digestive and Kidney Diseases scientists. Subsequent studies by investigators in the National Institute of Dental and Craniofacial Research (NIDCR), including four clinical trials, have provided a great deal of insight into the hormonal and bone abnormalities in patients and how to treat them, and how the disease evolves.

I appreciate your interest in current studies being done in the Craniofacial and Skeletal Diseases Branch of the NIDCR. These efforts are focused on three areas: 1) clinical management; 2) basic understanding of disease processes; and 3) therapeutic intervention. The NIDCR, in conjunction with the NIH Office of Rare Diseases, the FD Foundation, and the NIH Fogarty International Center, will be hosting an international workshop in the fall that will address all three areas. The NIDCR is continuing to prospectively study a large cohort of patients with MAS/FD for the purposes of further defining the spectrum and natural history of the disease, and examining the impact of evidence-based changes in clinical management. In the laboratory, studies are aimed at determining the consequences of the disease-causing mutations on cell functions, such as cell division, formation of different bone cell types, and cell life and death. These studies include collaborations across many other Institutes and Centers at NIH, including the NIH Chemical Genomics Center, and the National Institute on Deafness and Other Communication Disorders. Researchers are also investigating whether stem cells from unaffected bones of the patients could form normal bone in a cleaned-out FD lesion to prevent fracture and future disability. Alternatively, as the technology for inducible pluripotent stem (IPS) cells is developed on the NIH campus, investigators are hopeful that IPS cells isolated from the patient's own normal cells could also be used to form new bone.

Ultimately, we hope that pursuing all these lines of research will allow us to build on scientific opportunities for improved care and treatments for MAS/FD patients in the future. I appreciate your concern related to this important issue. Please do not hesitate to contact me again if you have additional questions or would like more information. I will also send a response to the co-signers of your letter.

Sincerely yours,

Francis S. Collins, M.D., Ph.D.
Director

List of Addressees

The Honorable Daniel K. Inouye
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The Honorable Jeffrey A. Merkley
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The Honorable Patty Murray
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The Honorable William D. Delahunt
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