Evaluating pharmaceutical treatments to replace surgery for bone overgrowth in Robinow Syndrome.

Motivation
- Robinow Syndrome is a rare genetic syndrome. The diagnostic features of Robinow Syndrome include shorter stature, a wider face and a prominent forehead.
- Some patients experience bone overgrowth that impinges on nerves and causes pain.
- The only treatment is neurosurgery to remove the bony overgrowths.

Project Description
- Using a mouse model (Prickle1 Beetjuice) and patient cells we will determine if FDA-approved drugs can modify the disease.

Context
- The only treatment option to relieve pain for these patients is surgically excise the bone overgrowth.
- Often the patients require several surgeries.
- Our analysis of the mouse model suggests that two pathways have altered signaling in bone cells. These pathways have FDA-approved drugs to modulate them.
- By identifying and testing FDA-approved drugs in our in vivo and in vitro model systems we hypothesize that we will be able to halt bone growth.

Potential Impact
- We hope to prevent the need for repeated neurosurgeries for patients with Robinow Syndrome.
- If the FDA-approved drugs modify the patient disease, then we will have identified a pharmaceutical treatment that can modify Robinow Syndrome.

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