Druggable targets for Robinow Syndrome

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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*^{Beetlejuice}) 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.

University of Pittsburgh

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

Project Deliverables

- Invention Disclosure filed with the Innovation Institute by 6 months.
- This project will provide the data for a competitive R01 application prior to the end of the funding period.
- Within 1-2 years, we expect to have submitted a high-impact paper with our results.

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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