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Decoding the Path to a Cure: Using zebrafish to Explore Craniometaphyseal Dysplasia

Nuwanthika Gimhani Wathuliyadde Ms
Western University, nwathuli@uwo.ca

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Decoding the Path to a Cure: Using zebrafish to Explore Craniometaphyseal Dysplasia

Craniometaphyseal Dysplasia is a rare human disorder characterized by abnormal bone mineralization, and affecting individuals of different ages with various symptoms. Severe conditions can lead to facial palsy, hearing loss and blindness and surgical recontouring of bone is currently the only available treatment. The disease itself is caused by impaired ANKH, a protein that transports inorganic pyrophosphate from intracellular to the extracellular environment, where inorganic pyrophosphate acts as an inhibitor for bone mineralization. Impaired ANKH transports less inorganic pyrophosphate extracellularly, leading to hyper-mineralization of the bone. My aim is to utilize zebrafish, a model organism for bone disease research, to generate an ANKH knockout line using the powerful gene editing tool, CRISPR-Cas9. These ANKH knockout animals will help to recapitulate the phenotypic abnormalities observed in humans afflicted with this disease, and more importantly will provide a platform for small molecule drug testing as potential therapeutic options.