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Caenorhabditis elegans as a rare disease characterization model

Daghar Hiba*, **Doyle James** and **Parker Alex J**
Alex Parker Lab, Canada

Rare congenital diseases are in urgent need of therapy. An animal such as the nematode caenorhabditis elegans may provide insights into the investigation in these diseases. Because of its known genome and the fact that many genes are highly conserved between

C. elegans and humans, it is possible to investigate the underlying pathways of these diseases. With up-to-date techniques such as CRIPR/CAS, we can generate patient- specific mutations. Our project aims concern the characterization of two rare congenital diseases: SRD5A3-CDG and Cori disease in C. elegans. These two diseases appear as pediatric congenital disease that are most likely underdiagnosed. Distinct and common phenotypes were identified to target them in further drug screen experiments. Interesting data show important motility impairment and neurodegeneration in C. elegans strains. We also present lifespan and progeny data and show how these will be relevant in further drug screening experiments. In this presentation, we will present our innovative tool in the modulization of rare diseases and how efficient it can be in such a field where the therapeutic need is urgent.

Biography

Daghar Hiba is a Master Student from Alex Parker Lab (CR-CHUM, Université de Montréal, Canada). Alex Parker Lab is specialized in working on genetic models for neurodegenerative diseases in C. elegans, stress response pathways in neurons and the discovery of drugs for neurodegeneration. Hiba's project, who is in the process of admission to the PHD, focus on rare congenital disease characterization in C. elegans. Promising data is leading us into a drug screening process.