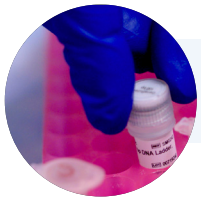
JORDAN'S SYNDROME: A JOURNEY TO A CURE

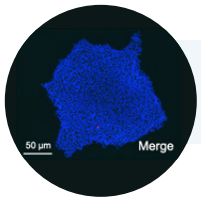
WHAT WE'VE DONE



CLINICAL PUBLICATION

Understand syndrome, symptoms, and prognosis → Publish

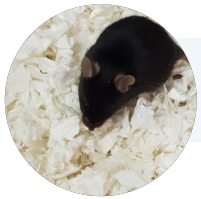
The growing number of individuals with Jordan's Syndrome enables a better understanding of the genetic mutations. The data collected from the families helps guide clinical care and future research efforts.



iPSCs CREATION

Collect human cells → iPSCs

Blood and skin samples were collected from individuals with Jordan's Syndrome and converted to neuronal cells or "brains in a dish".



MOUSE NATURAL HISTORY STUDY

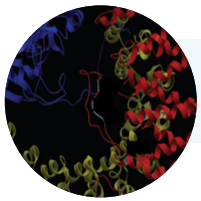
Create mice models → Use clinical data as a guide → Characterize behavior and brain development

Jordan's Syndrome mice models closely mirror the human condition. Characterizing the behaviors and taking a closer look at brain activity and function in the mice are critical steps to answering key questions such as ideal times for interventions.

NANOBODIES CREATION

Immunize alpacas with PPP2A subunit → Isolate antibodies and nanobodies

Gene specific antibodies and nanobodies were developed using alpacas as hosts.



3D MODEL DEVELOPMENT

Model PPP wild type protein structure → Determine location of the variants

Gene specific 3D protein structures were developed for the wild type and for each of the variants.

TOOLBOX

Outputs from stage 1 of the research that are key to moving the research effort forward.

The toolbox is now full!

Clinical Data

Induced Pluripotent Stem Cells (iPSCs)

Mice Models

Nanobodies

3D Protein Models

GOING FORWARD

TREATMENT

DRUG DESIGN

Perform high throughput drug screening → Determine promising drug compounds → Test on iPSCs → Perform mice clinical trials

USING:

→ **Identification of a drug for human clinical trials**

Jordan's Syndrome impacts the function of the brain. There is potential for a treatment that removes the effect of the genetic mutations. Finding the right drug can be an extensive process with a highly rewarding outcome.

CURE

GENETIC INTERVENTION

Perform gene modification using allelic specific oligonucleotide (ASO) → Test on iPSCs → Perform mice clinical trials

USING:

→ **Genetic intervention ready for human clinical trials**

ASO technology has the ability to turn off the misspelled copy of the gene with the potential of regaining typical function.