〈NGLY1 Deficiency Project: Development of therapeutic agents for rare hereditary diseases using iPS cells〉

Dr. Suzuki’s team is focusing on a deficiency in the NGLY1 gene that encodes for the de-N-glycosylating enzyme N-glycanase. They will develop innovative therapeutics for NGLY1 deficiency, a rare inherited disease that presently does not have any therapeutic options, through a combination of basic research findings, iPSC technology and a drug discovery platform.

Recent data suggested abnormalities in brain organoid developed from patient-derived iPSCs

- Many large neural tissues that have greatly expanded in wild type brain organoids but not in NGLY1-deficiency organoids (day 20).
- NGLY1-deficiency organoid which has failed to produce neuroepithelial buds, instead displaying extended cell processes consistent with direct neural differentiation.

Key Research Platform

- Patient-derived iPSCs
- NGLY1-KO mice

Clinical information

- w/ Physicians & Patients’ Foundation

Disease modelling

- Strategy for therapeutics
- Reproduction of disease-specific phenotype
- Assay system for drug screens
- Biomarkers for diagnosis & efficacy

Therapeutic options

- Existing & repositioning drugs
- Enzyme replacement
- Gene delivery

<Progress>