

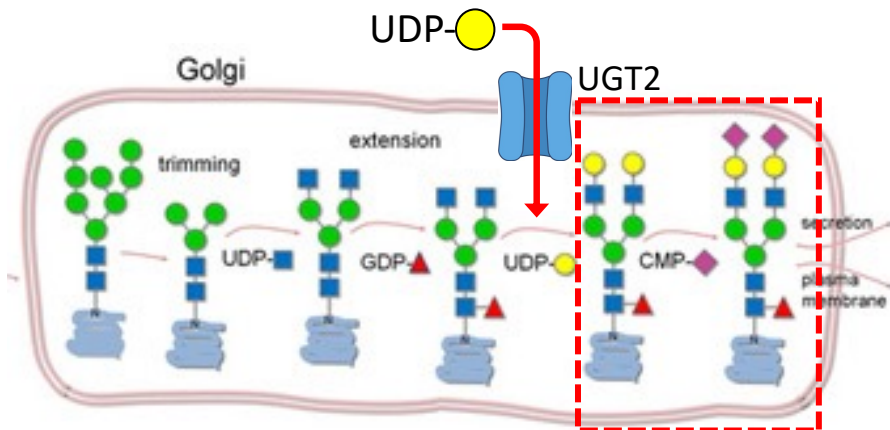
Establishing a human iPSC-derived neuron model of *SLC35A2*-associated epilepsy

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SLC35A2 (Solute carrier 35)

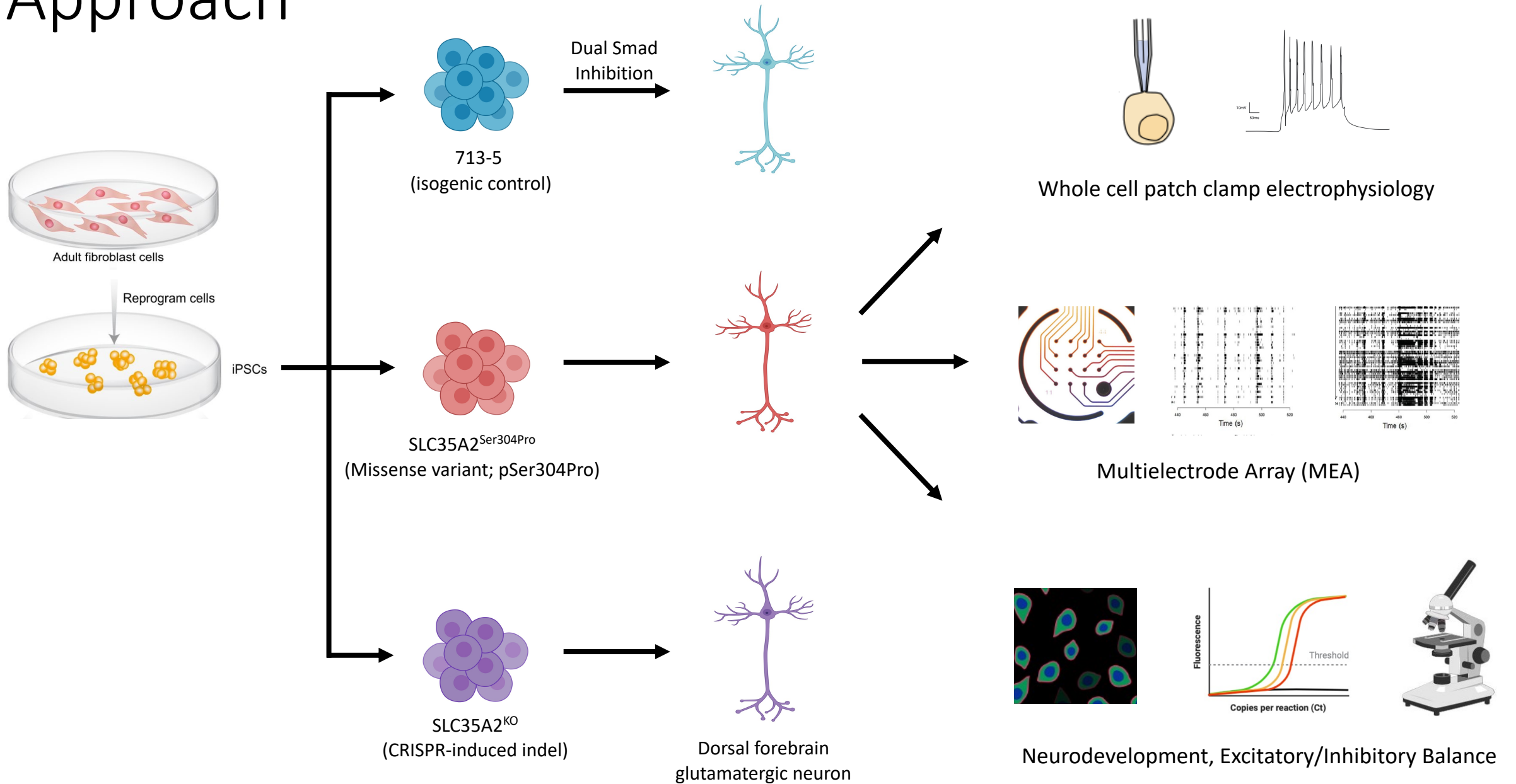
- Gene → Xp11.23
- Protein → UDP-galactose transporter (UGT2)
- Function → transports UDP-galactose from cytosol to Golgi lumen where it is used for the galactosylation of N-glycosylated proteins



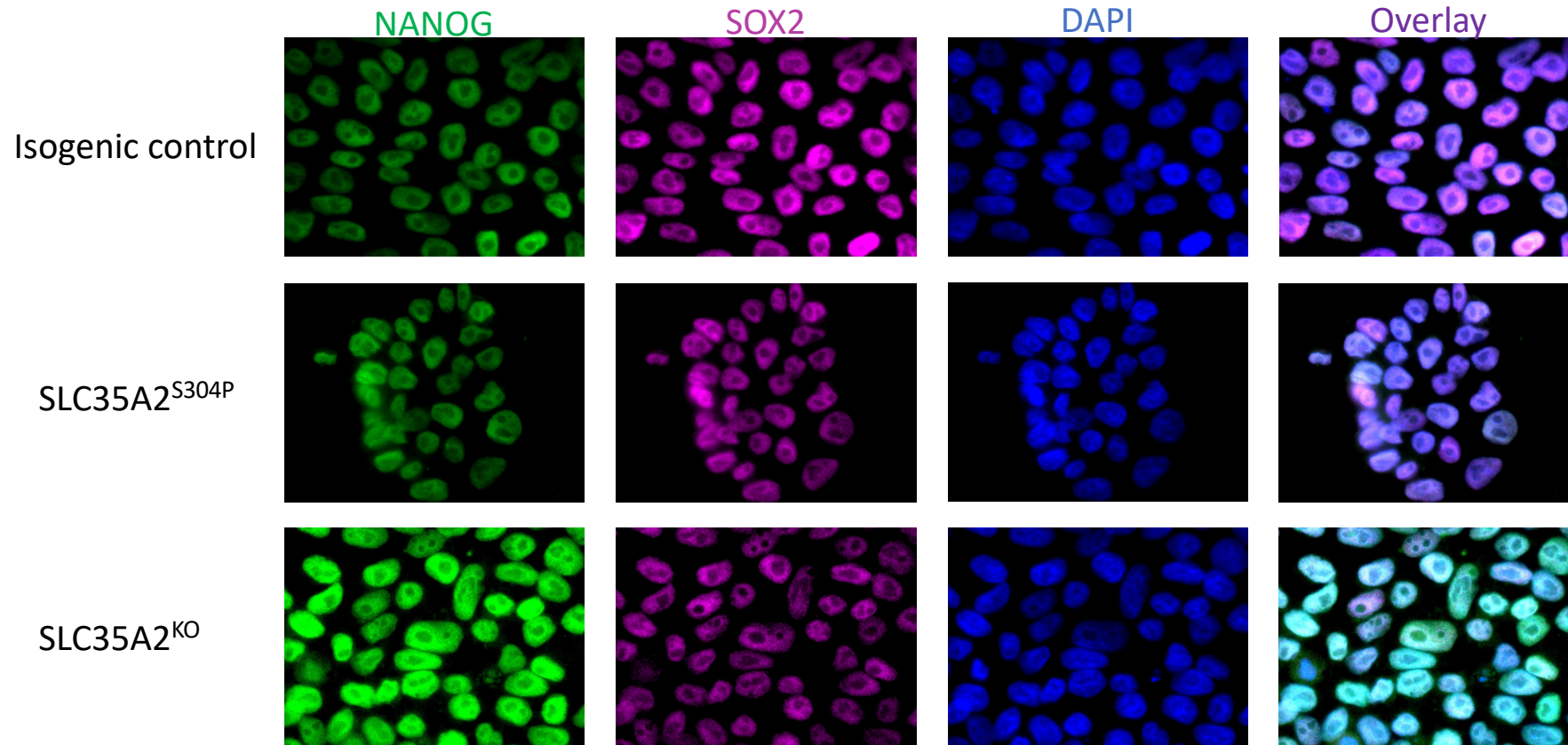
SLC35A2 and human disorders

- Congenital disorder of glycosylation
- X-linked developmental and epileptic encephalopathy
- Intractable neocortical epilepsy

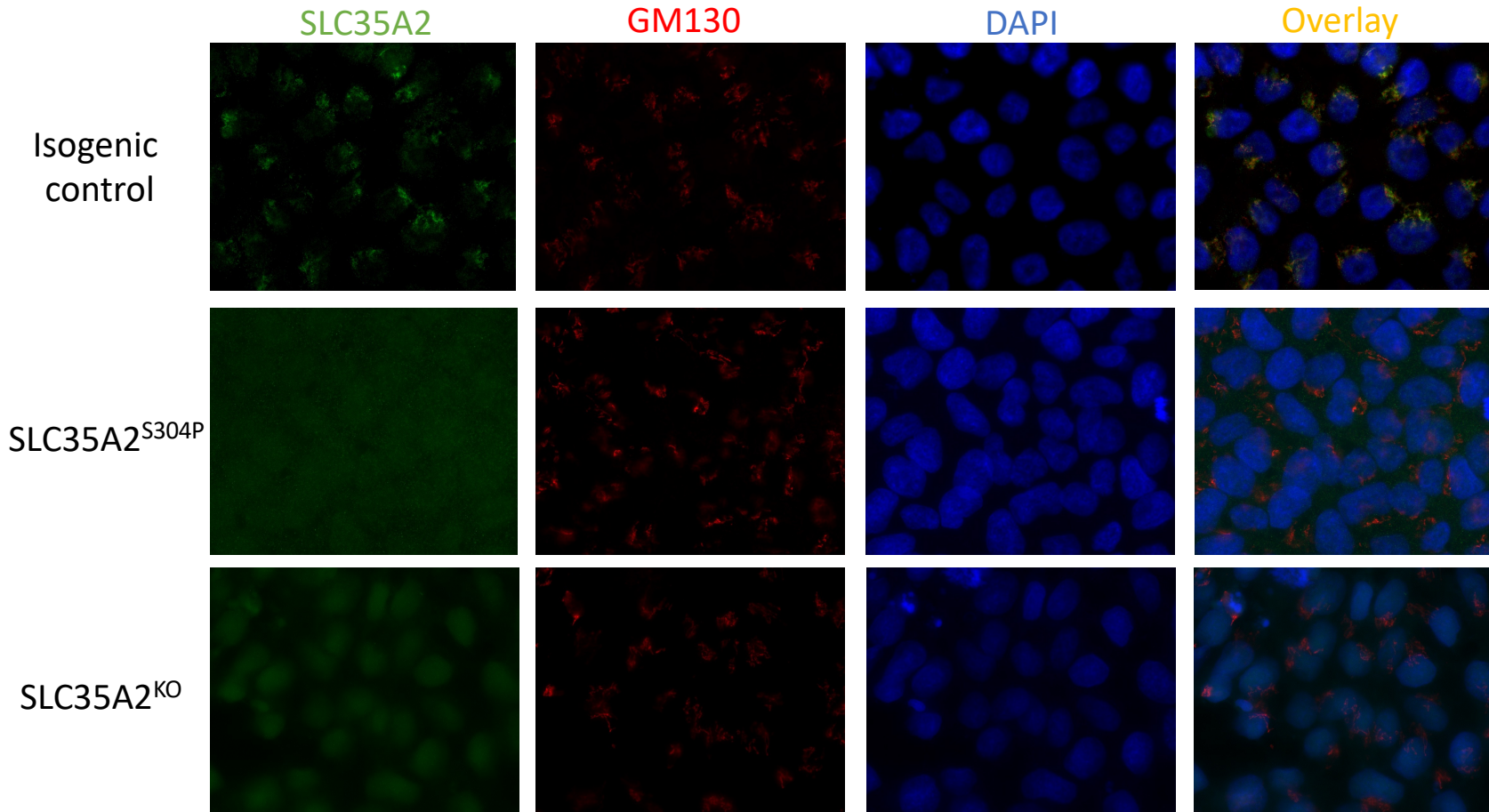
Approach



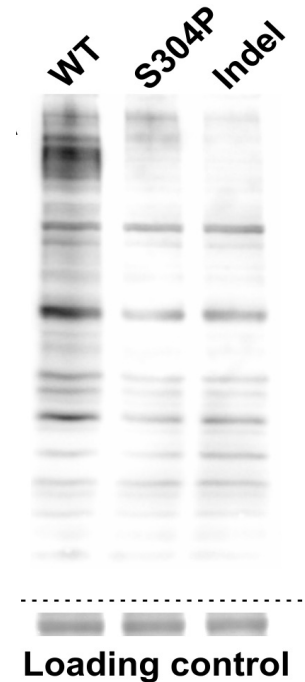
SLC35A2 CRISPR edited iPSCs retain pluripotent characteristics



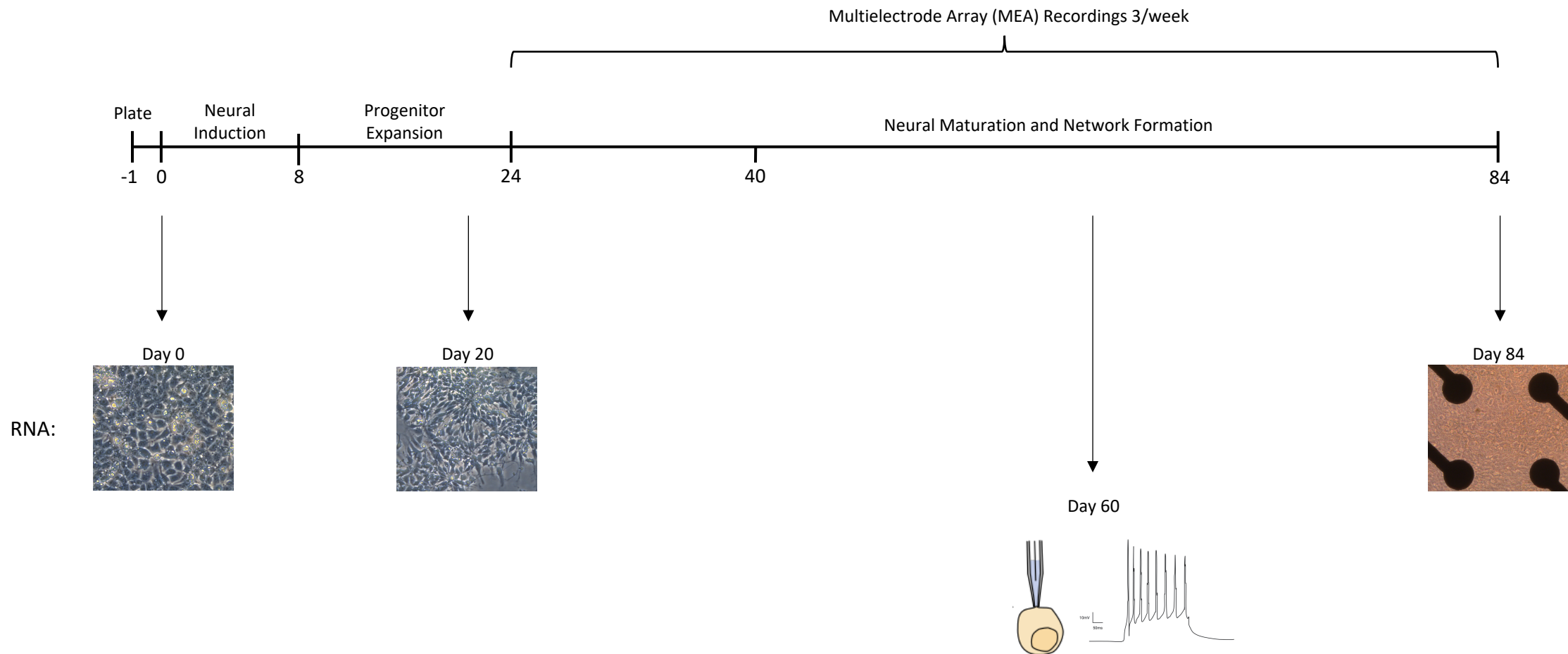
SLC35A2 variants encode LOF proteins resulting in reduced sialylated N-linked glycans



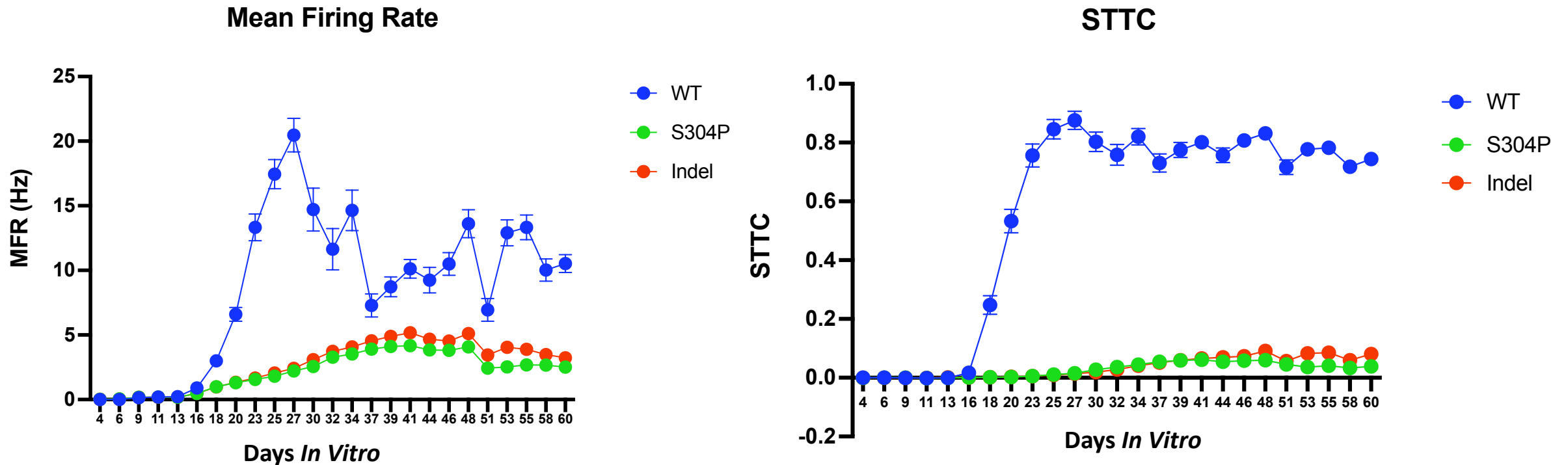
MAL-I Lectin Binding Assay



Experimental Timeline



SLC35A2 variant harboring neurons are hypoactive and generate asynchronous networks



*Days *in vitro* refers to the number of days plated on MEA from papain dissociation



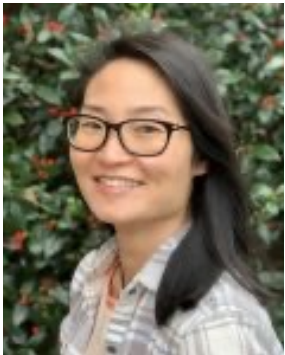
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