

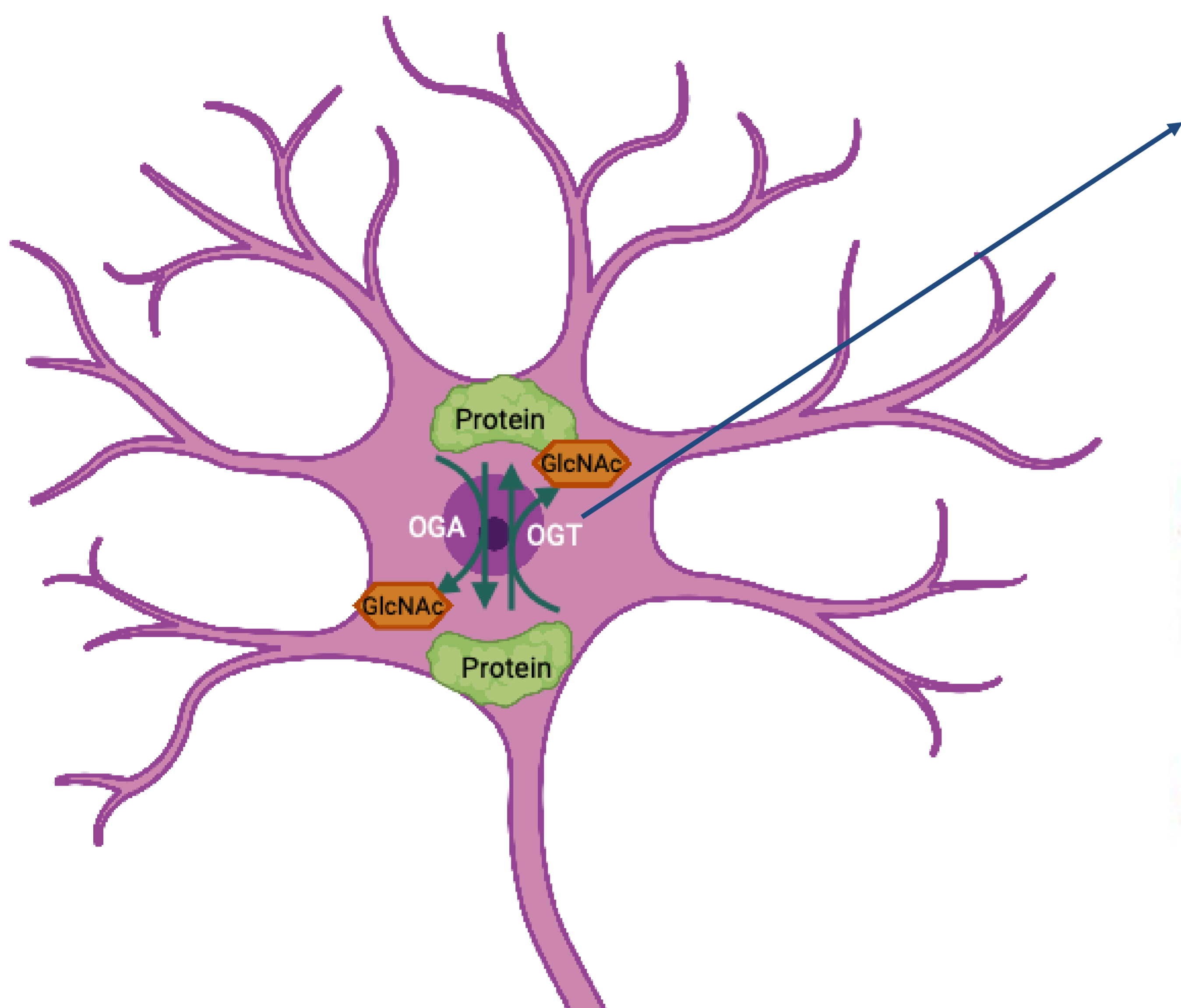


The role of dysfunctional post-translational modification to the development and progression of neurodevelopmental disorders.

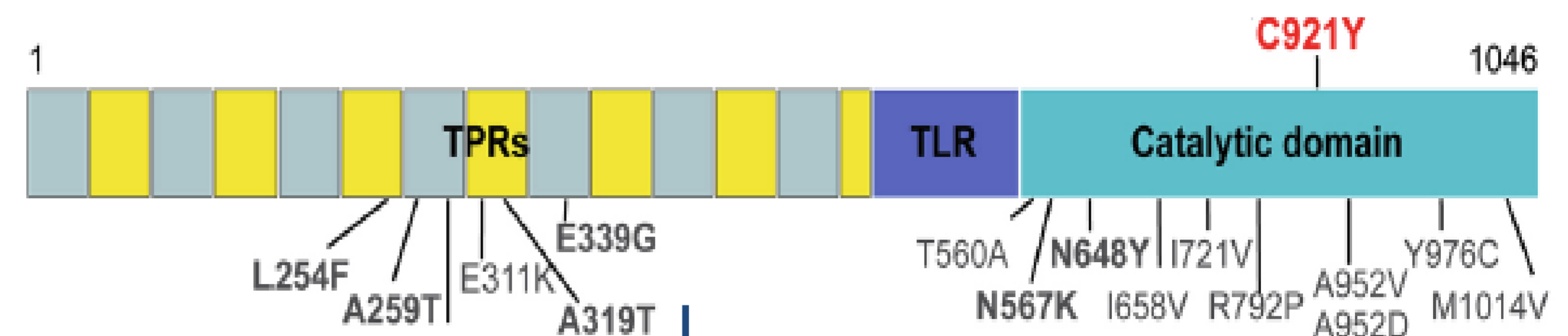
Daan van Aalten research group

Background:

O-GlcNAcylation



Mutations



Neurodevelopmental disorders

Brain

Intellectual disability, behavioural disorders/autism, brain malformations, psychomotor delay, epilepsy, dyskinesia

Eye

Strabismus, refractive errors

Language

Delay or absent development of speech

Face

Facial dysmorphism (microcephaly, thin upper lip, dolichocephaly, high-arched palate, etc.), café-au-lait spots

Locomotor system

Generalized hypotonia, hypertonia, joint hypermobility

Projects

1. What is the role of O-GlcNAcylation in neuronal development and maturation
2. What are consequences of OGT mutation in brain cells function?
3. What are the brain proteins that are O-GlcNAcylated?
4. Are some brain regions more vulnerable of impairment in O-GlcNAcylation than others and why?
5. Do some mutations give more severe phenotype than others?
6. Can we recreate patients symptoms in cell/animal models to study pathophysiology of the disorders?

Techniques

Experimental models:

- Stem cells
- Mouse and human primary neuronal/astrocytes/microglia
- oiu
- Mouse
- Artificial Intelligence

Techniques:

1. Cell culture
2. Microscopy
3. Immunoblotting
4. Flow cytometry
5. Proteomics
6. Animal behaviours
7. RNA sequencing
8. Viral transduction
9. Spatial transcriptomics
10. *In silico* analysis
11. Bioinformatics

Molekylærbiologi: Ja
Molekylær Medicin: Ja
Bioteknologi: Ja

novo
nordisk
fonden

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