The mechanisms of a new neurodevelopmental disorder

The new NNF Laureate-funded van Aalten lab @MBG,AU

What is O-GlcNAc?

- Post-translational modification
- Glycosylation
- Essential
- Conserved
- X-linked
- On >4K proteins
- Links with AD/PD

But what is its function?

What is OGT-CDG?

OGT missense mutations

"OGT-CDG"

Intellectual disability
Developmental delay
Epilepsy
Autism

PNAS (2019), EJHG (2020), Unpublished

Things we have done in the past

Structures
(e.g. EMBO J. 2006,2008)

Chemical tools
(e.g. Chem.Biol. 2010)

Development
(e.g. Nat.Chem.Biol. 2017)

Things we will do @MBG

Main aim: understand how OGT-missense mutations lead to intellectual disability

- Develop fly/mouse/ESC models
- Modulate using genetic/chemical tools
- Dissect through biochemistry/"omics"
- Understand through structural biology
- Interact with clinicians/patients

The lab @MBG from June 1st

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