

Technology Offer: Diagnosis and Treatment of Autism Spectrum Disorders Using a Deubiquitinase

Project Status

- **In vivo Study Complete**
- **Molecular Mechanisms Disclosed**

Benefits

- **Door-Opener to ASD Treatment**
- **Early Diagnosis**
- **High Medical Need**

Patents and Publications

- **European Patent Application Filed on 10.09.2021 by Universitätsmedizin der Johannes Gutenberg-Universität**
- **Pending PCT Application**
- **[Colombo et al., 2021, doi:10.1073/pnas.2110755118](https://doi.org/10.1073/pnas.2110755118)**

Offer

- **The Technology can be Licenced or Assigned**
- **Collaborations regarding further Development Welcome**

Scientists from Universitätsmedizin Mainz managed to identify the deubiquitinase CYLD as a key contributor to Autism Spectrum Disorder (ASD) –like traits. These findings pave the way towards solving two difficulties: Therapeutically modulating any manifestation of an ASD or associated neuropsychiatric disorders, and diagnosis of the mentioned conditions.

Autism spectrum disorders include neural developmental disorders characterized in particular by disturbances in social communication, stereotyped repetitive behaviors and interests, and manifest in early childhood. A high medical need is deduced from its prevalence: 1 % of the population is affected by ASD.

The Inventors established a mouse model lacking the murine CYLD gene. Detailed neurobiological analysis obtained the following results:

- 1) Loss of CYLD led to autism-like behaviors in mice. In particular, social communication was impaired. Increased repetitive behaviors occurred, and cognitive functions were impaired.
- 2) Functionally, the CYLD lack led to impaired synaptic transmission at defined contact sites in the CA1 region of the hippocampus.
- 3) Morphologically, the CYLD absence resulted in decreased lengths of dendrites and a reduced number of dendritic spines.

Thus, the inventors demonstrate for the first time that loss of the CYLD gene is associated with behavioral and functional morphological and molecular biochemical changes linked to ASD hallmarks.

Therefore, decreased CYLD levels can be used as a diagnostic tool, aiming to be used before ASD manifestation in the early childhood or beyond. Most importantly, agents that increase the CYLD activity could be used to treat autism and intelligence impairment.

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The Universitätsmedizin Mainz team would look forward to cooperate with commercial partners to transfer the invention towards industry and market exploitation.