Exploring the role of the immune system in a model of Helsmoortel-van der Aa syndrome

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Loss-function mutations in the ADNP gene are linked to the Helsmoortel-Van der Aa Syndrome (HVDAS), a complex neurological disorder characterized by autism and intellectual disability. Adnp mutant mice recapitulate key hallmark of ASD including learning deficit, increased anxiety and repetitive behavior. ADNP is involved in brain development, neuroprotection and modulation of the immune system. Despite a clear link exists between ADNP and the immune system, to date the exact role of ADNP in immune cell is unknown, and we ignore the contribution of the immune system to the progression or severity of the HVDAS.

 A better understanding of the immune changes associated with ADNP deficiency may provide clues to pathological processes and allow to identify biological markers that enable early diagnosis and treatment of ASD. In this project, we will explore the role of ADNP in T cells function, to this end will perform in depth characterization of the molecular and cellular signature acquired by T cells and microglia cell populations in the mutant mice.