Lay Summary

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Cellular and genetic pathomechanisms of central pontine myelinolysis

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Central pontine myelinolysis (CPM) and Alexander's diseases (AxD) are rare neurological disorders with similar symptoms. One of these symptoms is the loss of special coating on nerve cells called myelin. With only a handful of cases diagnosed every year, it is difficult to conduct research into how these diseases work. We think that CPM and AxD have separate processes and can result in different outcomes. However, similarities in disease symptoms prompted us to ask a question whether both diseases have similar underlying processes. To test this, we will analyse a particular type of brain cells called glia (astrocytes and microglia in particular) in patients with AxD and CPM and compare them to control cases. We hope that this project will help us to help our understanding into internal mechanisms of these rare and devastating disorders. We request brain samples from CPM patients for comparison with available samples from AxD patients and healthy individuals. This will mean that we can sequence a gene encoding a glia marker protein called glial fibrillary acidic protein (GFAP). This might be a possible gene which might be mutated in CPM patients and could potentially cause CPM as well as AxD.