TSC patient iPSC brain-on-a-chip to investigate severity of Epilepsy

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Discordance of TSC severity

In general, patients with Tuberous sclerosis complex (TSC) caused by loss of TSC2 endure more severe symptoms then when caused by absence of TSC1. However, if the same mutation is inherited from parent to child the symptoms may vary greatly between them. To find underlying causes for this discrepancy, we utilize induced pluripotent stem cells (IPSCs) that have been reprogrammed from blood cells, donated by such discordant patients. The huge benefit of this technique is that we can transform patient-derived IPSCs into human neurons and/or astrocytes.

With this approach we want to model epilepsy with a brain-on-the-chip to investigate neuronal function and to decipher factors that cause the variation in severity. These might help predict the severity of TSC patients early on, assess potential treatment options, and in the long run improve disease severity and quality of life.