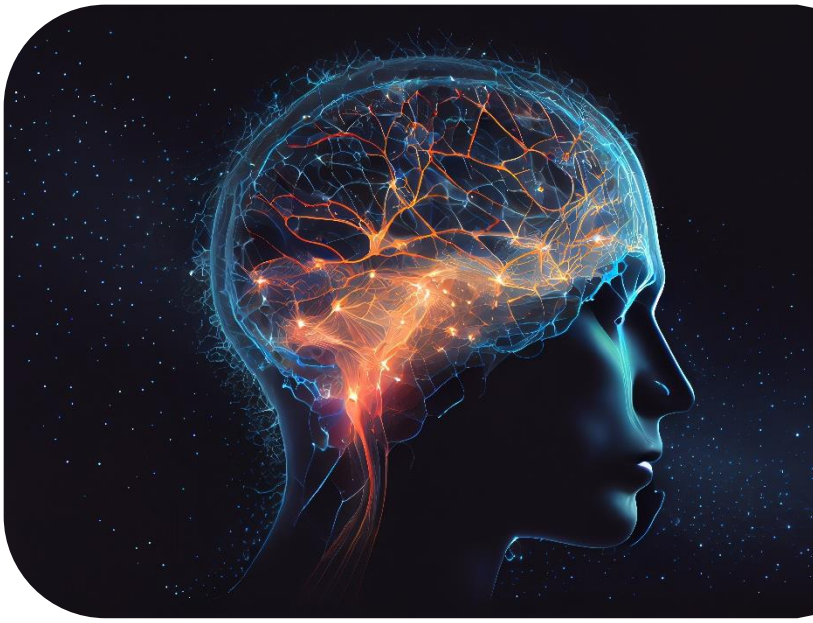
>> Autism spectrum disorder

Autism spectrum disorder (ASD) is a neurodevelopmental disorder that can affect communication, social interaction, and behavior. While some genetic and environmental risk factors have been linked to ASD, causes typically remain complex and unknown. Recently, improved disease models and access to patient induced pluripotent stem cells (iPSC) are enabling scientists to better characterize ASD phenotypes *in vitro*. The Maestro platform offers sensitive functional analysis of these phenotypes, both to gain a better understanding of pathogenic mechanisms as well as insight into the efficacy of potential therapeutics.



Learn how the Maestro can support your ASD research with **these selected publications:**

MYT1L haploinsufficiency in human neurons and mice causes autism-associated phenotypes that can be reversed by genetic and pharmacologic intervention

Bettina Weigel, Jana F. Tegethoff, et al. Molecular Psychiatry. (2023)

MYT1L mutations are strongly associated with ASD diagnosis, but the underlying mechanisms are not fully understood. Researchers explore these mechanisms and potential therapeutic interventions using primary rodent neurons and human stem cell-derived neurons.

Highlights:

- MYT1L-mutant mice exhibit ASD phenotypes, including hyperactivity, social deficits, and delayed neurogenesis.
- MEA provides functional measurement of hyperactivity in both haploinsufficient MYT1L hESC-derived and primary mouse neurons.
- Both genetic correction via MYT1L overexpression and pharmacological treatment reduce hyperexcitability in MYT1L-mutant neurons.

Read more >>

Elevated levels of FMRP-target MAP1B impair human and mouse neuronal development and mouse social behaviors via autophagy pathway

Yu Guo, Qiping Dong, et al. Nature Communications. (2023)

Fragile X syndrome (FXS), caused by deficiency in Fragile X messenger ribonucleoprotein 1 (FMRP), is the leading genetic contributor to autism spectrum disorder (ASD), but the underlying mechanisms remain unclear. Researchers characterize FRP-deficient neurons and evaluate a potential therapeutic.

Highlights:

- FMRP deficiency leads to increased MAP1B levels and ASD-like behavior in adult mice.
- Maestro is used to characterize patient iPSC-derived neurons, finding increased MAP1B levels and hyperexcitability.
- Genetic correction via FMRP knockdown or pharmacological treatment with rapamycin reduces hyperexcitability, suggesting a therapeutic target and potential candidate.

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Transcription Factor 4 loss-of-function is associated with deficits in progenitor proliferation and cortical neuron content

Fabio Papes, Antonio P. Camargo, et al. Nature Communications. (2022)

Transcription Factor 4 (TCF4) has been linked to autism spectrum disorder, schizophrenia, and other neuropsychiatric disorders, yet the relationship between TCF4 and neuronal function is poorly understood. Researchers use ASD patient iPSCs to create neural organoids as a model of TCF4 dysfunction.

Highlights:

- Neural organoids generated from Pitt-Hopkins syndrome patients exhibit reduced proliferation and differentiation, resulting in developmental deficits and decreased size.
- Using the Maestro, TCF4-mutant organoids are found to be less active than wild-type organoids.
- Neural activity is rescued via CRISPR/Cas9-based genetic correction.

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Species-specific FMRP regulation of RACK1 is critical for prenatal cortical development

Minjie Shen, Carissa L. Sirois, et al. Neuron. (2023)

Previous research has associated FMRP deficiency with mitochondrial dysfunction, but it remains unclear how this relates to Fragile X syndrome (FXS) and autism spectrum disorder (ASD). Researchers explore this relationship using human fetal cortical slices and patient iPSC-derived neurons.

Highlights:

- FMRP regulates gene expression during development, contributing to mitochondrial and neuronal dysfunction.
- FMRP-mutant neurons are hyperexcitable, as measured via MEA.
- Enhancement of mitochondrial function rescues FXS-associated deficits, suggesting mitochondrial function as a therapeutic target.

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