



TSA's Research Strategy

The TSA is committed to funding research in line with its six research priorities, which are consistent with the charity's five-year strategy to reach more people and drive more research. Our vision is a world where TSC and its effects are conquered.

Our research priorities from 2019-2023 are:

Earlier diagnosis, including:

- Improving foetal ultrasound strategies
- Using neonatal EEG in those at risk to spot early signs of epilepsy
- Genetic testing through universal maternal blood tests and/or universal blood drop testing for neonates

Early risk assessment, including:

- Targeted genetic testing for families with known risks
- Genetic analysis combined with manifestations (genotype: phenotype correlations)
- Using natural history registers and databases to calculate predicted future severity of condition

Basic science and pre-clinical work, including:

- Lab-based research to understand molecular mechanisms
- Lab-based research to identify new therapeutic targets and establish effectiveness in a pre-clinical setting
- Stimulating initial stage research of TSC-gene modifying therapy in pre-clinical studies

Prevention, including:

- Trials of mTOR inhibitors and other therapies for newly diagnosed infants and older people aimed at preventing kidney damage, preventing lung damage (LAM) and earlier control of infantile spasms and other manifestations of TSC
- Diagnosis and early control of infantile spasms
- Tools to alert parents of infantile spasms and how to report them to TSC clinics
- Approaches to lowering the risk of developing neuropsychiatric disorders

The Tuberous Sclerosis Association
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Registered address: The TSA, Unit 56, 1 Emma Street, London, E2 9FP
Charity number: 1039549 (England & Wales), SC042780 (Scotland)



Research into new and available therapies, including:

- Optimising therapy for kidney disease, skin disease, LAM and epilepsy (better and more tailored treatments)
- Promoting methods for the early identification of TSC-Associated Neuropsychiatric Disorders (TAND), measuring unmet need and improving treatments for each aspect of TAND.

Social and service research, including:

- Looking at what is really affecting people's lives and how to support them (for example, isolation, inability to work, education)
- Understanding best practice in delivering support and care from the TSA

Every scientist and research project which the TSA funds must be focused on the potential benefits for people living with TSC, their families and carers. Applications should therefore familiarise themselves with the above research strategy.

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