



Accelerating Progress:

**RESEARCH STRATEGY
2025-2028**

To provide **help** for today
and a **cure** for tomorrow

We are delighted to introduce the TSA's new research strategy.

Since its launch, the Tuberous Sclerosis Association (TSA) has established itself as a world leader in tuberous sclerosis complex (TSC) research, driving forward both scientific understanding and meaningful progress that makes a real difference to the lives of people affected by TSC.

This strategy marks an important milestone. It has been shaped through extensive consultation with researchers, clinicians, and, most importantly, the wider TSC community. We are deeply grateful to everyone who has contributed their time, insight, and experience to help ensure the strategy reflects both our ambition and the practical realities of advancing research in TSC.

While research into any rare condition can be challenging and complex, our commitment to enable world-leading research into all areas of TSC remains steadfast, and this strategy provides a roadmap for action and collaboration.

We hope this strategy inspires confidence and enthusiasm among our partners, supporters, and the broader research community. Together, by working in partnership and sharing knowledge, we can continue to accelerate progress and improve outcomes for everyone affected by TSC.

Thank you again to everyone who has contributed to this important work. Your support and dedication is what makes the TSA a driving force in rare disease research.

Joint Chief Executives

**Dr Pooja Takhar and
Luke Langlands**





We envision a future where TSC is better understood, diagnosed earlier, and treated more effectively.

Our research aims to uncover the underlying biology, identify new therapeutic opportunities, and translate discoveries into meaningful improvements for people living with TSC and their families.

Collaboration is central to this vision. By connecting researchers, clinicians, and the TSC community, we aim to accelerate progress, share knowledge, and ensure that research addresses the needs that matter most.

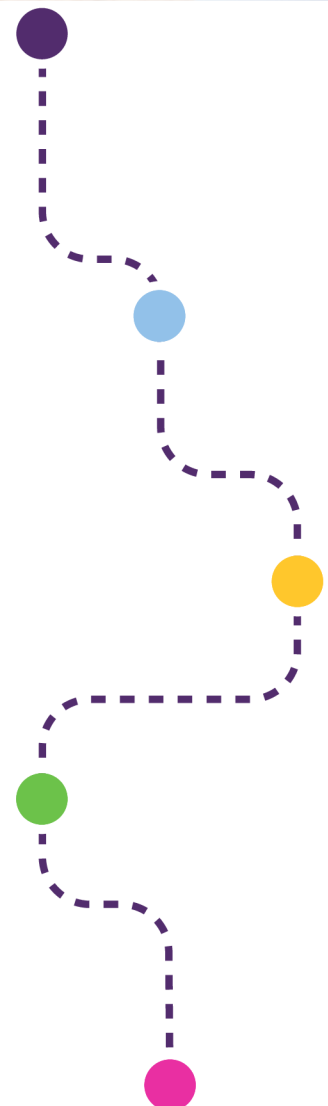
Ultimately, our goal is to **drive innovation that changes lives** — turning scientific discovery into real-world impact and creating a future where people living with TSC have better outcomes, treatments, and hope



A research strategy is essential to build on the significant advances our work has already helped to achieve. Over the years, research supported and facilitated by the TSA has contributed to earlier and more accurate diagnosis, a better understanding of the genetic and biological mechanisms underlying TSC, and the development of new treatments that are improving outcomes for people living with TSC.

Despite this progress, many challenges remain. There are still delays in diagnosis for some people, limited treatment options for certain manifestations of TSC, and a need for more innovative therapies that address every aspect of TSC. The refreshed strategy allows us to focus resources, strengthen collaboration across the research community, and ensure that studies are aligned with the most pressing needs of everyone affected by TSC.

By setting clear priorities, we can accelerate discoveries and translate them into meaningful improvements in care.



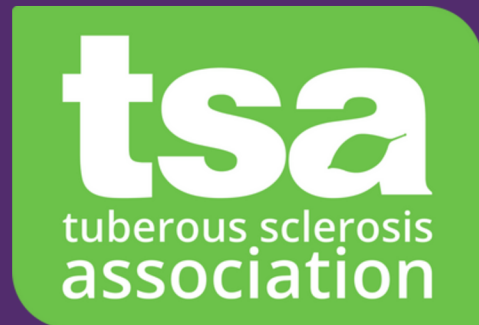


Research priorities are vital because they give us a clear focus, helping us direct resources towards the areas that can make the biggest difference for people with TSC.

They provide a framework for the projects we fund, the partnerships we build, and the opportunities we pursue, ensuring that every step is guided by the needs of the TSC community and informed by scientific evidence. By setting out clear priorities, we can accelerate progress, avoid duplication, and make sure that the research we support delivers real benefits for individuals and families affected by TSC.

We will support research to:

- 1) Understand the lifelong impact of TSC**
- 2) Bring about effective medical treatments**
- 3) Optimise care with focus on early diagnosis and monitoring**



Lifetime impact

RESEARCH
PRIORITY ONE

To provide **help** for today
and a **cure** for tomorrow



Our goal is to deepen understanding of the lifelong impact of TSC on individuals and families.

This means building a clearer picture of how TSC affects health, development, and wellbeing at every stage of life, and recognising the wide variation in experiences across the community. By exploring the social, educational, psychological, and economic consequences of TSC, we aim to highlight unmet needs, inform better support, and drive research that improves quality of life throughout the lifespan.

Through research we will

- **Understand adulthood in TSC**

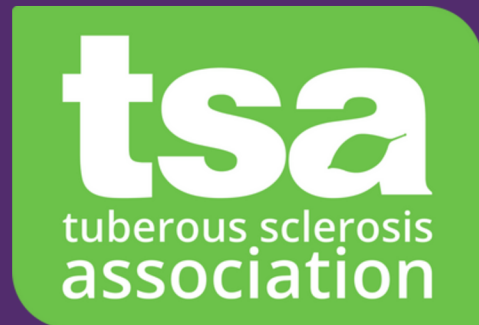
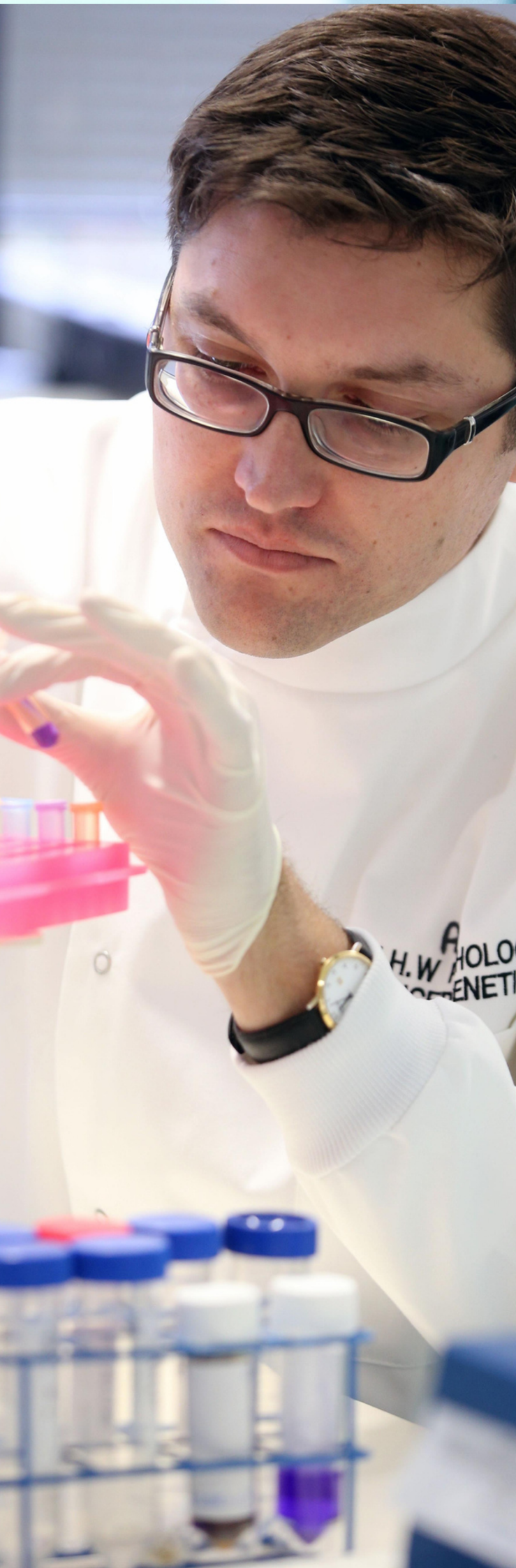
By researching how TSC affects individuals across the adult lifespan, and identifying the biological, psychological, and social factors that influence health, wellbeing, and long-term outcomes.

- **Investigate hormonal stages**

By exploring how key hormonal stages — such as puberty, pregnancy, and menopause — influence progression, symptoms, and wellbeing in people with TSC.

- **Identify early life predictors**

By using tools such as natural history databases to uncover behaviours, symptoms, and biomarkers in early life that shape long-term outcomes and quality of life for people with TSC.



Treatments

RESEARCH PRIORITY TWO

To provide **help** for today
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Our goal is to ensure that people with TSC benefit from the best possible treatments.

This means improving how we manage TSC-Associated Neuropsychiatric Disorders (TAND) and epilepsy expanding access to effective pharmacological and non-pharmacological interventions, and strengthening understanding of how current treatments work in real life. By optimising existing options and driving forward new approaches, we aim to improve both day-to-day management and long-term outcomes for everyone affected by TSC.

Through research we will

- **Improve outcomes for TSC-Associated Neuropsychiatric Disorders (TAND)**

By exploring pharmacological, behavioural, educational, and psychosocial interventions to improve outcomes for people living with the TAND.

- **Optimise epilepsy therapies**

By making the best use of existing and emerging therapies to improve epilepsy management in TSC, reducing seizures and improving quality of life.

- **Better understand treatments in practice**

By building knowledge about the effectiveness, tolerability, and interactions of treatments already used in TSC, ensuring real-world evidence guides better care and outcomes.

- **Explore gene-based therapies**

By investigating the potential of gene-based approaches to treat or modify TSC, laying the groundwork for future breakthroughs and transformative treatments.



Optimising care

RESEARCH
PRIORITY THREE

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Our goal is to ensure that everyone with TSC has access to fair, consistent, and effective care throughout their lives.

This means understanding and addressing the barriers that prevent people from receiving the services they need, refining the ways we monitor TSC across the lifespan, and embracing digital technologies that can transform care and improve quality of life. By optimising care pathways and making the best use of innovation, we aim to deliver more timely interventions, better management, and greater equity for all those affected by TSC.

Through research we will

- **Understand barriers to care**

By identifying the challenges that prevent fair and consistent service delivery for people with TSC, and developing evidence-based solutions to improve access and equity.

- **Optimise surveillance**

By defining the most effective ways to monitor TSC across the lifespan, including the right approaches, tools, and intervals to deliver the best outcomes.

- **Harness digital technologies**

By using digital tools and innovations to transform how TSC is monitored and managed, while supporting independence and improving quality of life.



To achieve our research vision, we know it is not enough to set priorities, we must be clear about how we will deliver them.

These enablers are the essential things we will focus on to make our ambitions a reality. They provide the structure, support, and momentum that will allow us to accelerate progress, strengthen research, and ensure that everything we do is grounded in collaboration and the lived experience of people with TSC.



Collaborating for impact

Partnership is at the heart of progress. We will work with researchers, clinicians, funders, industry, and international networks to combine expertise and resources, ensuring that research into TSC achieves more than any organisation could deliver alone.

Embedding lived experience

The voices of people affected by TSC are essential in shaping research. From setting priorities to designing studies and participating in trials, we will ensure that lived experience guides and strengthens research at every step through the TSC Research Volunteer Network.

Driving innovation

We will embrace new ideas, technologies, and approaches that have the potential to transform TSC research. By supporting bold and creative science, and by building capacity in the research community, we will accelerate progress towards our research vision.



To deliver our research strategy, the TSA is committed to funding only the highest quality research with the greatest potential to improve the lives of people affected by TSC.

All funding decisions are made through a rigorous peer review process, in line with the AMRC's six principles of expert review: proportionality, independence, diversity, rotation, impartiality, and transparency. Each application is reviewed by scientific and medical experts, people with lived experience of TSC, and our research funding committees, with final approval from the TSA Board of Trustees.

Between 2025 and 2028, we will invest in research and research capacity by supporting projects that range from early exploratory studies to early career researchers. This includes flexible funding for promising and underfunded areas, ensuring we can respond quickly to new opportunities. We support researchers across universities, hospitals, and institutions in the UK and internationally, from early-career scientists to established leaders.

Partnerships remain central to our approach. By working with other charities, funders, and industry, we can extend our reach and amplify impact. Our commitment also goes beyond awarding grants: we will support researchers by encouraging collaboration, sharing findings, and ensuring that the results of TSA-funded research are openly available for the benefit of the global TSC community.

THANK YOU

RESEARCH STRATEGY



We are deeply grateful to the community members, researchers, and clinicians who generously shared their experiences, insights, and expertise to shape this strategy. Your contributions ensure that our priorities reflect the realities of living with TSC, the opportunities in science, and the shared ambition to improve lives.

This strategy is stronger because of you.





We're here for everyone affected by TSC only thanks to the amazing people and groups who kindly provide us with regular and one-off donations

Help us to continue to be here for you and others, now and the future:

tuberous-sclerosis.org/donate

Switchboard:
0300 222 5737
admin@tuberous-sclerosis.org

TSA Support Line:
0808 801 0700
support@tuberous-sclerosis.org
Webchat (tuberous-sclerosis.org)

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tuberous sclerosis
association

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