Medical Xpress

Survey on tuberous sclerosis complex finds fragmented care, lack of information, scarcity of support

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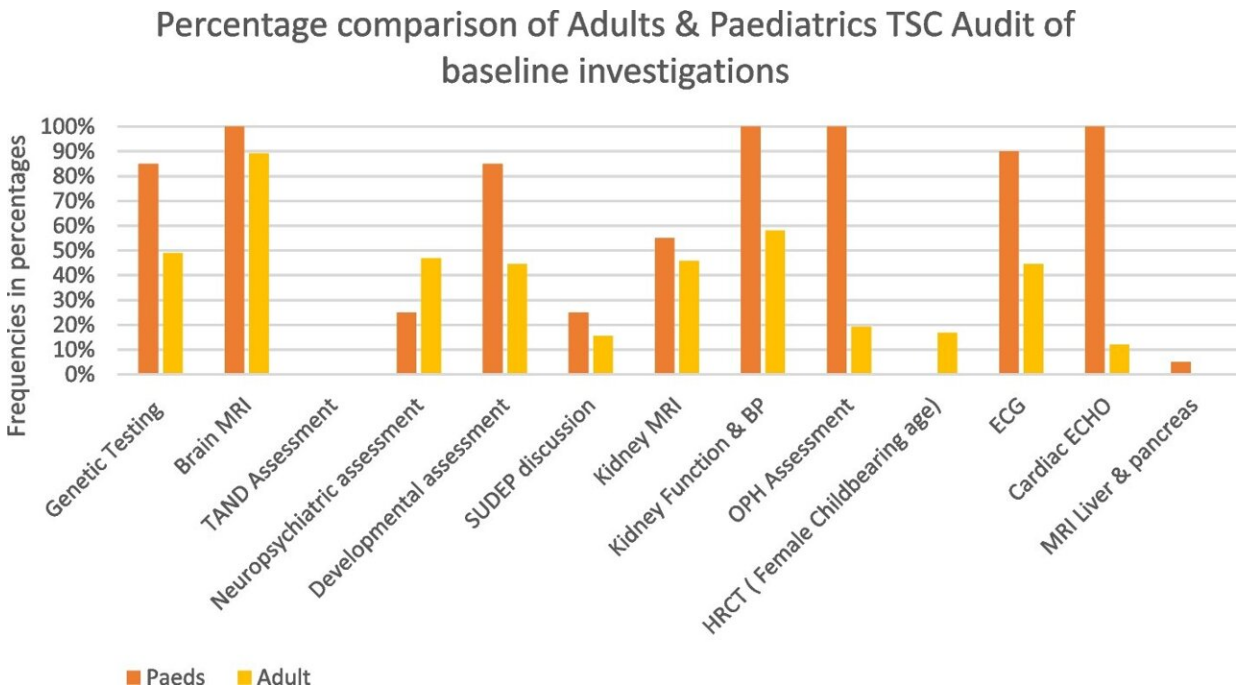

Credit: *Journal of Rare Diseases* (2024). DOI: 10.1007/s44162-024-00049-8

Researchers call on policymakers, decision-makers, and "those who hold the purse strings" to listen to the evidence and take action to enable individuals with TSC to receive the internationally recommended coordinated care they require to live full lives.

Tuberous sclerosis complex (TSC) is a rare genetic disease (approximate 1:6,000 birth incidence) with a wide variability of physical and neuropsychiatric symptoms. Patients require lifelong care from multiple health care specialists.

But what is known of TSC in the Republic of Ireland? Up to now, very little, despite the fact that there are an estimated 600 patients living with the disease. Today, the findings of the first national clinical audit of baseline TSC care, of a group of adult and pediatric patients attending epilepsy services, carried out by Trinity College Dublin researchers, have been published in the *Journal of Rare Diseases*.

The paucity of information surrounding the illness, including the health care of this group, what care is being provided, if any, the lived experience of this group and the needs of the health care professionals who provide their care, led the Trinity team to undertake this research.

Researchers found care to be fragmented and lacking a formal TSC clinic or network to provide the internationally recommended TSC care for this multiorgan complex disease.

Coordination of the many investigations and consultations required to manage TSC frequently falls on the shoulders of the patients, families and caregivers to navigate their care and health care system.

Researchers say this is not conducive with the International or UK TSC recommendations, where lifelong, personalized, multidisciplinary care, by a coordinated team of TSC experts is recommended. Nor is it in line with the WHO guidance on safe systems of care to improve patient safety and reduce available harm.

Key findings

- Care was shown to be fragmented with no centralized TSC care coordination as per the UK recommendations.
- Numbers (adult and children) attending epilepsy services were lower than expected. 135 patients were attended compared to approx. 400 patients that would have been expected to attend.
- Many baseline investigations were completed especially in pediatric services. However, separate referrals were required to access services and treatments even within the same hospital, resulting in fragmented care.
- All specialties required for the care of TSC patients were available in the ROI, e.g., neurology, nephrology and investigative technology such as MRI were available. However, long waiting times and no clear feedback systems resulted in fragmented uncoordinated care.

Key recommendations

The establishment of a TSC center of excellence in the ROI, with the recruitment of a TSC coordinator.

Coordinated care, ideally in a center of excellence, would facilitate health care professionals with TSC expertise to provide lifelong personalized care to TSC patients and support to their HCP colleagues. There would be an amplification in TSC awareness, increased patient safety and outcomes, improved patient and provider satisfaction and a reduction in health care costs.

Researchers note that an advanced nurse practitioner passionate about TSC has already begun to coordinate TSC patient care of those who are attending St James Hospital in Dublin. The team report that this has been lifechanging for these patients and the service needs to expand.

The research team highlighted that in recent years, important progress

has been made in the management and control of TSC with the emergence of new pharmaceutical treatments. It is vital people receive an early diagnosis and the appropriate treatment.

Mary Vasseghi, Ph.D. candidate, School of Medicine Trinity College Dublin and FutureNeuro, the study lead author, said, "The understanding of the mechanism of TSC has changed and critically, improved the trajectory and outcomes for individuals living with TSC .

"The development of targeted pharmaceutical treatments and the establishment of International Clinical Recommendations for the management of this complex disease has greatly contributed to this progress.

"It is vital individuals receive an early diagnosis and ongoing appropriate surveillance and management by a coordinated team with TSC expertise-for life. We must provide coordinated integrated care for these patients. It is life-saving and life-changing.

As a parent of someone living with TSC and a new researcher in this field, I feel privileged to have this unique perspective."

Claire Behan, Ph.D. candidate, Academic Unit of Neurology, Trinity College Dublin and Registered Advanced Nurse Practitioner Epilepsy, Neurology Department, St. James's Hospital and co-author, said,

"Tuberous sclerosis is a rare genetic disorder that significantly impacts individuals and families in Ireland. Raising awareness through research like this is essential in supporting those affected. The backbone of this research is the network of advanced nurse practitioners in epilepsy across Ireland, highlighting the impact of interdisciplinary, collaborative research.

"Based on the findings from our initial audit, we began implementing changes at a local level. However, with these latest insights, it has become clear that these improvements should be extended and standardized at a national level.

"Policymakers, decision-makers, and those who hold the purse strings must listen to the evidence before them and take action. We need to empower parents to focus on their families and enable individuals with TSC to live their lives fully—free from the burden of navigating complex care systems.

"It's time to stand with our international colleagues in delivering a seamless, inclusive, and efficient service for people in Ireland with TSC, grounded in evidence and ultimately saving costs. The responsibility to act is now."

More information: M. Vasseghi et al, Widespread service fragmentation for patients and families with tuberous sclerosis complex (TSC) in the Republic of Ireland, *Journal of Rare Diseases* (2024). DOI: 10.1007/s44162-024-00049-8

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