Expert recommendations for the provision of coordinated care for TSC

Just like in Australia, the rarity and complexity of TSC presents a number of challenges to the implementation of best practice health care in the UK. Treatment and follow-up are frequently reported to be fragmented, disjointed and suboptimal.

This excellent paper, which is well worth reading in its entirety, is written by an eminent team of TSC expert health professionals in the UK. It discusses the need to improve TSC management to ensure people with TSC have early access to appropriate treatment and preventive measures—both to minimise the long-term effects of the condition, where possible, and to support individuals and their families.

Given the range of organ systems affected by TSC, its management and treatment requires coordination across a number of medical specialties over a person's lifetime. The authors propose a comprehensive, holistic model of care to manage a range of manifestations which require management from a wide range of specialties.

The current model of TSC health care in the UK

Currently in the UK, there are 16 specialist TSC clinics. These have often been founded by enthusiastic and committed clinicians, but are frequently inadequately funded. And, significantly and sadly, it is still the case that most people living with TSC in the UK are not currently managed within these specialist centres. And, just like in Australia, the transition from paediatric to adult services can be a particularly challenging time for people with TSC in the UK.

There are, however, some beacons of hope. In Wales, a specialist TSC clinic has been established through a partnership between a pharmaceutical company and the National Health Service. However, it is currently awaiting the development of a fully sustainable commissioning model. And, in Northern Ireland, a TSC clinic has been running since 1995 which directly reviews the majority of people living with TSC in that region.

Hub and spoke model with tailored care

The UK team proposes a 'hub and spoke' model of care with a central network of TSC clinics/centres coordinated by specialists, and supported by a regional network of clinicians who offer access to a comprehensive set of TSC-related specialist services.

In accordance with the International Guidelines on TSC Surveillance and Management, they suggest the treatment and long-term surveillance needs of TSC should be determined based on the extent of disease at baseline and should be tailored to the individual.

The authors propose that specialists should work collaboratively with individuals, their families and their community doctors (general practitioners or GPs) to provide support and advice and a pathway for dealing with problems that need specialist care. Since holistic care of people living with TSC requires input from many different specialties, treatment should be discussed within the regional network by a multidisciplinary team, with the aim of ensuring that each TSC individual and their family have a tailored care plan to manage current disease manifestations and surveillance for future TSC manifestations.

The paper recommends that specialist TSC services should ensure the following:

- **Diagnosis:** People with TSC are identified by clinical evaluation and/or genetic testing
- **Surveillance:** Provision of multi-disciplinary evaluation through alignment with regional genetic services (for genetic counselling) and with other clinical specialties to ensure access to appropriate care for all
- **Treatment:** The appropriate access and use of TSC therapies
- **Safe transition from paediatric to adult care**
- **Information and support:** Collaboration with individuals/family and other organisations to provide access to TSC-specific information
- **Research:** Facilitate individuals and their families becoming involved in relevant research projects.

The authors propose that regional TSC clinics should be responsible for the diagnosis of TSC and the provision of routine care and support for people affected by TSC and their families. Regional clinics should be supported by a dedicated TSC specialist coordinator who has responsibility for coordinating the service, ensuring timely surveillance and coordinating care between different specialist services, developing individualised plans for follow-up and ensuring continuity of care for young people transitioning to adulthood.

They suggest that linking these regional clinics with TSC support organisations (like TSA in Australia) is vital to ensuring that individuals and their families receive comprehensive support.

The authors also propose that the regional clinics are in an ideal position to gather data to monitor needs locally and facilitate future TSC research.

Core services

To allow regional TSC clinics to fulfil this role, it is proposed that they need to offer, or have access to, a range of core services, including:

- Genetic testing and genetic counselling
- Neurology and neuroimaging
- Nephrology, urology, general and interventional radiology services
- Clinical psychology, psychiatry, and developmental paediatrics
- Collaboration with support organisations
- Collaboration with community physicians (GPs).

In the proposed model, the regional centres should have access to the necessary facilities to cater for the specific needs of people affected by TSC (such as brain and renal imaging performed under general anaesthetic) and access to additional specialist support services (such as Dermatology, Respiratory, Cardiology, Neuropsychiatry and Obstetrics/Gynaecology).

The authors recommend that, where regional centres are unable to provide a core service, there should be a clear pathway through which that service can be accessed.

Holistic care

In the hub and spoke model, as well as facilitating access to the appropriate services, the regional centres should be responsible for ensuring holistic care for individuals with TSC and their families and provision of supportive care, including referral for individualised education plans, genetic counselling for family members and ongoing support from an organisation like TSA in Australia.

The authors highlight the need to ensure that individuals are offered appropriate, regular surveillance and timely follow-up. They propose that people with multiple complications of TSC should attend joint clinics or have the monitoring of different manifestations performed in a single session, e.g. combined surveillance/monitoring of SEGA and renal AML through a coordinated MRI scan of both brain and renal tract, particularly where a general anaesthetic is required to achieve the imaging.

In terms of the transition for people moving from paediatric to adult services – a time when they are often lost to follow up – the authors propose that the TSC regional centres should ensure the service is aligned with guidelines on how to manage transitional care, with bespoke plans drawn up where necessary.

Collaboration

As well as highlighting the need for specialists to work collaboratively with people affected by TSC, their families and their GPs, collaboration with local/community services such as community paediatricians and mental health and between paediatric and adult centres (if they are not co-located services) is seen to be critical to success of the model.

Whilst the holistic management of TSC is often managed by a hospital specialist, the primary physician responsible for other aspects of care is usually the GP, and therefore collaboration and good communication between all players is essential to ensure the delivery of coordinated care.

Continuous improvement

Finally, the paper suggests services should be regularly reviewed to help to identify any potential opportunities for improved efficiency and ensure that people with TSC are consistently screened and treated according to best practice.

The authors also acknowledge that optimal management of TSC is a field of active research and new recommendations will continue to be made.

Conclusion

The model of care recommended by this team of UK experts involves centralised specialist TSC hubs, with routine management coordinated centrally and undertaken in regional TSC networks. The hub and spoke model would form a coordinated care network that will also provide a structure to facilitate the education of health care professionals and affected families and facilitate TSC research. TSA supports further discussion and action locally and globally on this important topic.

Annear, N.M., Appleton, R., Bassi, Z., Bhatt, R., Bolton, P., Crawford, P., Crowe, A., Tossi, M., Elmslie, F., Finlay, E. and Gale, D.P., 2019. Tuberous sclerosis complex (TSC): expert recommendations for provision of coordinated care. Frontiers in neurology, 10, p.1116.