

2017 年结节性硬化症（TSC）的 31 个事实

1. **COMPREHENSIVE** - Tuberous sclerosis complex (TSC) is a genetic disorder that causes tumors to form in vital organs, primarily the brain, eyes, heart, kidneys, liver, lung and skin.

概况- 结节性硬化症是一种遗传病，可导致脑、眼睛、心脏、肾脏、肝脏、肺脏和皮肤等重要器官发生肿瘤。

2. **GENETIC** - Tuberous sclerosis complex (TSC) is caused by a mutation in either the TSC1 or TSC2 genes.

遗传 - 结节性硬化症由 TSC1 或 TSC2 基因发生致病性突变引起。

3. **EARLY** - Many babies with tuberous sclerosis complex (TSC) are now diagnosed before or soon after birth when rhabdomyomas (a type of heart tumor) are detected during routine ultrasounds.

早期 - 许多结节性硬化症患儿在出生前或出生后可通过常规心脏超声检查发现心脏横纹肌瘤（一种心脏肿瘤）而得以确诊。

4. **DIVERSE** - Tuberous sclerosis complex (TSC) affects everyone differently, even identical twins.

多样性 - 结节性硬化症患者表现不尽相同，即使是同卵双胞胎。

5. **ANYBODY** - Tuberous sclerosis complex (TSC) shows no gender bias and occurs in all races and ethnic groups.

任何人 - 结节性硬化症的发生无性别差异，可见于所有种族。

6. **COMMON** - More common than Lou Gehrig's disease (ALS) or cystic fibrosis, tuberous sclerosis complex (TSC) is far less known.

普遍 - 尽管结节性硬化症比肌萎缩侧索硬化（渐冻人）或囊性纤维化常见，它却鲜为人知。

7. **SCIENCE** - Everolimus and rapamycin, which are used to treat some aspects of tuberous sclerosis complex (TSC), were developed thanks to scientists studying

bacteria in the soil of Easter Island.

科学 - 治疗结节性硬化症的依维莫司和雷帕霉素得益于科学家们对源自复活节岛土壤中细菌的研究。

8. RARE – TSC is one of the 7000 rare diseases in the world.

罕见病 - 结节性硬化症是 7000 种罕见病的一种。

9. INTERDISCIPLINARY - A team approach to health care, with multiple specialists working together, can benefit people with tuberous sclerosis complex (TSC). Most people with TSC do not have access to this style of care.

学科之间 - 多学科的专家协作诊疗可让结节性硬化症患者受益，但多数患者尚未惠及。

10. NEUROLOGICAL - At least half of people with tuberous sclerosis complex (TSC) have normal intellectual ability, but they may still experience difficulties with neurological manifestations that affect their daily life functioning.

神经 - 尽管一半以上结节性硬化症患者的智力正常，但他们的神经系统异常仍然可影响其日常生活。

11. DIAGNOSIS – Thanks to new sequencing generation, genetic diagnosis of TSC was substantially improved and should be done for any people diagnosed with TSC.

诊断 - 新一代测序技术显著提高了结节性硬化症的基因诊断率，所有的结节性硬化症病人应该做基因检测。

12. IMPACT - Tuberous sclerosis complex (TSC) affects more than 200, 000 individuals in China and many more carers, families and friends who live with the impact of the disease.

影响 - 在中国，结节性硬化症罹及 20 多万人，并牵涉到众多的护理人员 and 亲朋好友。

13. DOCTORS – More than 10 specialists are sometimes needed for a TSC patient.

医生 - 有时候，一个结节性硬化症病人需要 10 个以上的专家。

14. RESEARCH – Tuberous sclerosis complex (TSC) research contributes to our understanding of cancer, autism, epilepsy and other diseases.

研究 - 结节性硬化症的研究惠及了我们对肿瘤、自闭症、癫痫和其它疾病的了解。

15. WORLDWIDE - Around 1 in 6,000 babies born are affected by tuberous sclerosis complex (TSC). Worldwide, more than 1 million people have TSC.

全世界 - 每 6000 新生儿中有一个结节性硬化症患者，全世界有 100 万以上的结节性硬化症患者。

16. HAPPENING - Every 20 minutes somewhere in the world, a child is born with tuberous sclerosis complex (TSC).

发生 - 全世界每隔 20 分钟就有一个结节性硬化症患者降生。

17. ASSOCIATION - All TSC associations around the world are working together to improve life for people living with TSC. In China, it is TSCChina (www.tscchina.org).

病友组织 - 中国北京蝴蝶结结节性硬化症罕见病关爱中心 (www.tscchina.org) 正与世界各地的病人组织共同努力，改善结节性硬化症患者的生活。

18. FAMILY - Quality of life for partners and siblings could be substantially affected by TSC.

家庭 - 结节性硬化症可严重影响伴侣和至亲的生活质量。

19. CAUSAL - Tuberous sclerosis complex (TSC) is the most common genetic disorder associated with epilepsy and autism.

病因 - 结节性硬化症是最常见伴有癫痫和自闭症的遗传性疾病。

20. LESIONS - Angiofibromas are raised red bumps that grow on the faces of people with tuberous sclerosis complex (TSC).

病变 - 面部血管纤维瘤是结节性硬化症患者面部红色突起的小包块。

21. EPILEPSY - Seizures occur in approximately 85% of people with tuberous sclerosis complex (TSC), making epilepsy the most common brain manifestation of TSC.

癫痫 - 约 85% 的结节性硬化症患者有癫痫发作，因此癫痫是结节性硬化症患者最常见的脑部症状。

22. SEIZURES - Infantile spasms are a common type of seizure in babies with tuberous sclerosis complex (TSC). Vigabatrin is recommended by TSC experts as first line treatment, where available, to control these seizures.

癫痫发作 - 婴儿痉挛症是结节性硬化症患者常见的癫痫发作类型。若条件允许，TSC 专家推荐氨己烯酸（喜保宁）作为一线治疗药物控制癫痫发作。

23. INCURABLE - There is no cure for tuberous sclerosis complex (TSC).

不可根治 - 结节性硬化症尚无法治愈。

24. GLOBAL - TSC International (TSCi) tuberous sclerosis complex (TSC) organisations work with local health care professionals to improve the care given to the 1 million people around the world living with TSC.

全球 - 国际结节性硬化症组织（www.tscinternational.org, TSCi）与各地医护人员合作，为全世界 100 万 TSC 患者提供更好的关爱。

25. OPTIONS - Research in tuberous sclerosis complex (TSC) has already provided new treatment options for people with TSC. However, many people around the world do not have access to these medicines or the recommended surveillance for TSC.

选择 - 结节性硬化症的研究已经为结节性硬化症患者提供了新的治疗选择。然而，世界各地的很多患者无法获得这些药物或无法得到推荐的随访。

26. SPECTRUM - Almost half of people with tuberous sclerosis complex (TSC) will be diagnosed with an autism spectrum disorder.

疾病谱系 - 大约一半的结节性硬化症患者有自闭症相关障碍。

27. KIDNEYS - Up to 90% of people with tuberous sclerosis complex (TSC) will have tumors in their kidneys called angiomyolipomas.

肾脏 - 高达 90% 的结节性硬化症患者有肾脏血管肌脂瘤。

28. BURDEN - Tuberous sclerosis complex (TSC) can be associated with considerable disease burden for patients and caregivers.

负担 - 结节性硬化症可对病人和看护人造成沉重的负担。

29. BEHAVIOUR - Behavioural difficulties in tuberous sclerosis complex (TSC) can include anxiety, depressed mood, overactivity, restlessness, aggression, temper tantrums, self-injury, social communication difficulties and sleep problems.

行为 - 结节性硬化症行为异常包括焦虑、抑郁、兴奋、坐立不安、攻击、发脾气、自残、社交困难和睡眠障碍。

30. COUNSELING - All people with tuberous sclerosis complex (TSC) should receive genetic counseling when diagnosed.

咨询 - 所有诊断为结节性硬化症的患者需要得到遗传咨询。

31. LUNGS - Lymphangiomyomatosis (LAM) can occur in the lungs of people with tuberous sclerosis complex (TSC), almost exclusively in females. June 1 is worldwide LAM awareness day.

肺脏 - 淋巴管平滑肌瘤病（LAM）几乎仅出现在女性结节性硬化症患者的肺部，6月1日是世界LAM关注日。