

2017 年結節性硬化症 (TSC) 的 31 個事實

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.

概況 - 結節性硬化症是一種遺傳病，可導致腦、眼睛、心臟、腎臟、肝臟、肺臟和皮膚等重要器官發生腫瘤。

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

遺傳 - 結節性硬化症由 TSC1 或 TSC2 基因發生致病性突變引起。

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 - 許多結節性硬化症患者兒在出生前或出生後可通過常規心臟超聲檢查發現心臟橫紋肌瘤（一種心臟腫瘤）而得以確診。

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

多樣性 - 結節性硬化症患者表現不盡相同，即使是同卵雙胞胎。

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

任何人 - 結節性硬化症的發生無性別差異，可見於所有種族。

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

普遍 - 儘管結節性硬化症比肌萎縮側索硬化（漸凍人）或囊性纖維化常見，它卻鮮為人知。

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.

科學 - 治療結節性硬化症的依維莫司和雷帕黴素得益于科學家們對源自復活節島土壤中細菌的研究。

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

罕見病 - 結節性硬化症是 7000 種罕見病的一種。

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.

學科之間 - 多學科的專家協作診療可讓結節性硬化症患者受益，但多數患者尚未惠及。

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.

神經 - 儘管一半以上結節性硬化症患者的智力正常，但他們的神經系統異常仍然可影響其日常生活。

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

diagnosed with TSC.

診斷 - 新一代測序技術顯著提高了結節性硬化症的基因診斷率，所有的結節性硬化症病人應該做基因檢測。

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.

影響 - 在中國，結節性硬化症罹及 20 多萬人群，並牽涉到眾多的護理人員和親朋好友。

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

醫生 - 有時候，一個結節性硬化症病人需要 10 個以上的專家。

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

研究 - 結節性硬化症的研究惠及了我們對腫瘤、自閉症、癲癇和其它疾病的瞭解。

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 新生兒中有一個結節性硬化症患兒，全世界有 100 萬以上的結節性硬化症患者。

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

發生 - 全世界每隔 20 分鐘就有一個結節性硬化症患者降生。

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).

病友組織 - 中國北京蝴蝶結結節性硬化症罕見病關愛中心 (www.tscchina.org) 正與世界各地的病人組織共同努力，改善結節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 - 結節性硬化症可嚴重影響伴侶和至親的生活品質。

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

病因 - 結節性硬化症是最常見伴有癲癇和自閉症的遺傳性疾病。

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

病變 - 面部血管纖維瘤是結節性硬化症患者面部紅色突起的小包塊。

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

癲癇 - 約 85% 的結節性硬化症患者有癲癇發作，因此癲癇是結節性硬化症患者最常見的腦部症狀。

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.

癲癇發作 - 嬰兒痙攣症是結節性硬化症患者常見的癲癇發作類型。若條件允許，TSC 專家推薦氨己烯酸（喜保甯）作為一線治療藥物控制癲癇發作。

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

不可根治 - 結節性硬化症尚無法治癒。

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 - 國際結節性硬化症組織（www.tscinternational.org, TSCI）與各地醫護人員合作，為全世界 100 萬 TSC 患者提供更好的關愛。

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.

選擇 - 結節性硬化症的研究已經為結節性硬化症患者提供了新的治療選擇。然而，世界各地的很多患者無法獲得這些藥物或無法得到推薦的隨訪。

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

疾病譜系 - 大約一半的結節性硬化症患者有自閉症相關障礙。

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

腎臟 - 高達 90%的結節性硬化症患者有腎臟血管肌脂瘤。

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

負擔 - 結節性硬化症可對病人和看護人造成沉重的負擔。

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.

行為 - 結節性硬化症行為異常包括焦慮、抑鬱、興奮、坐立不安、攻擊、發脾氣、自殘、社交困難和睡眠障礙。

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

諮詢 - 所有診斷為結節性硬化症的患者需要得到遺傳諮詢。

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.

肺臟 - 淋巴管平滑肌瘤病 (LAM) 幾乎僅出現在女性結節性硬化症患者的肺部。6月1日是世界 LAM 關注日。

