

Lesão ungueal: um olhar além da queixa dermatológica

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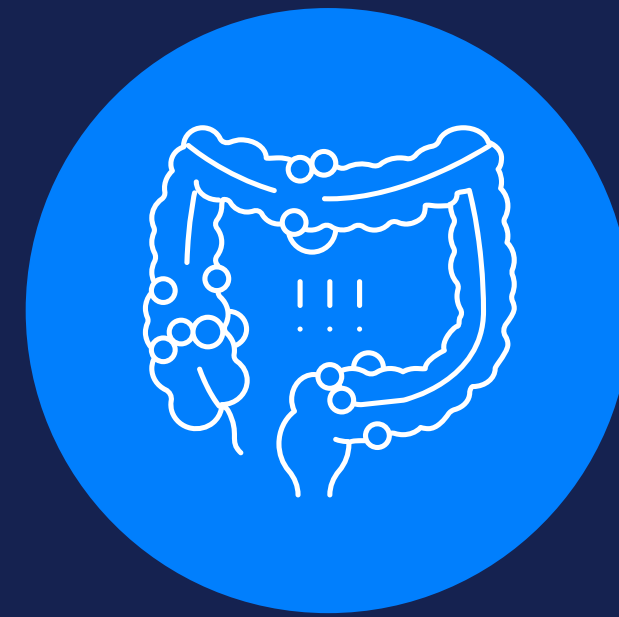
Caso Clínico



**Masculino
21 anos**



**"Lesão na unha do pé
há mais de um ano"**



**Doença de Crohn
diagnóstico há 2 anos**



Hipótesis diagnósticas

Fibroqueratoma periungueal adquirido?

Fibromixoma acral superficial?

Pólipo Fibroepitelial?

Tumor de Koenen?





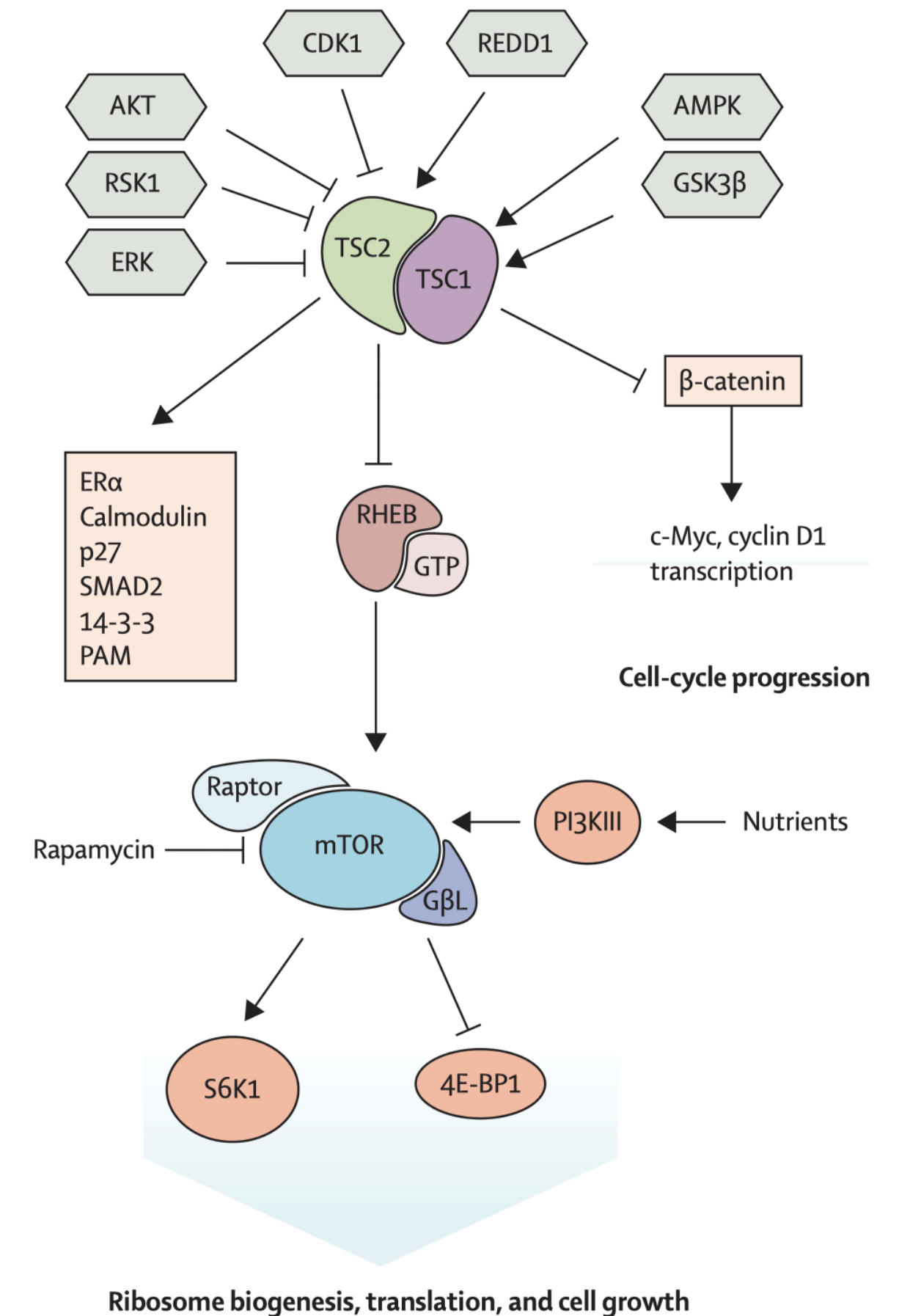




Esclerose Tuberosa

Esclerose Tuberosa

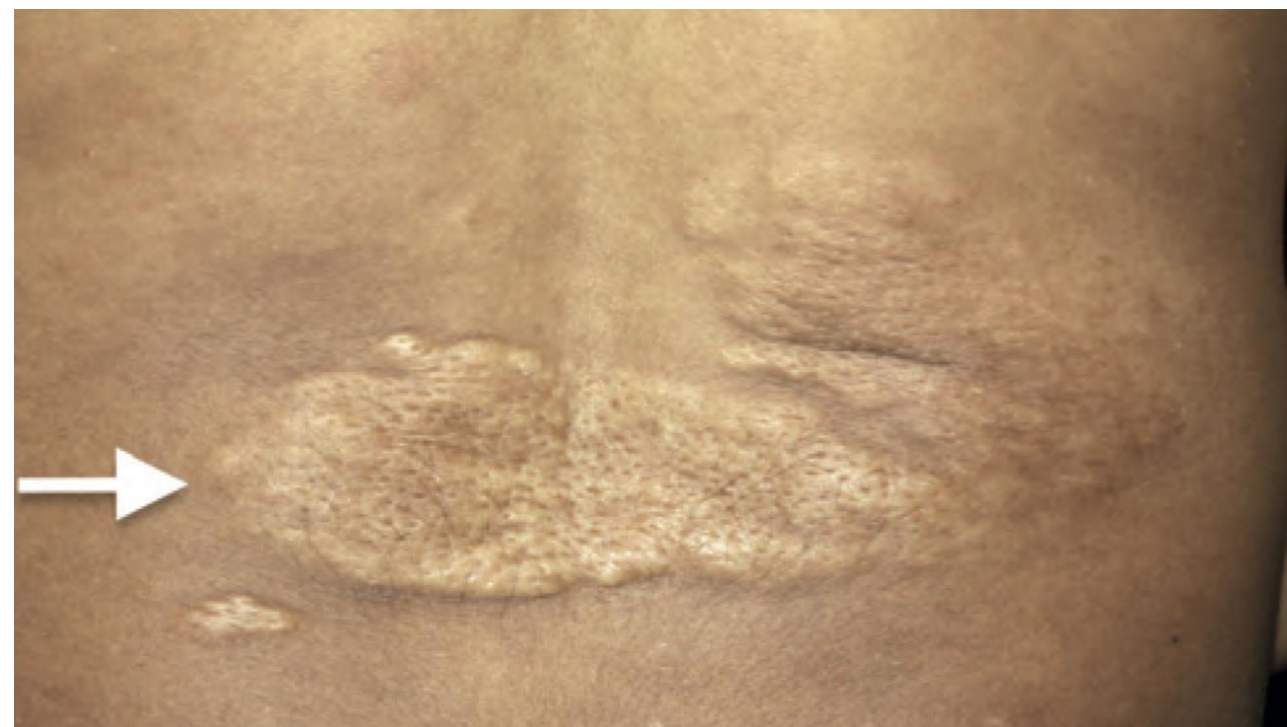
- Doença genética multissistêmica
- Genes TSC1 ou TSC2
- Hamartina e Tuberina (via mTOR)
- Desenvolvimento de tumores benignos (hamartomas)
- 80% mutações novas



Manifestações mucocutâneas são os achados mais comuns



Máculas hipocrômicas (80%)



Placa de Shagreen



Tumor de Koenen



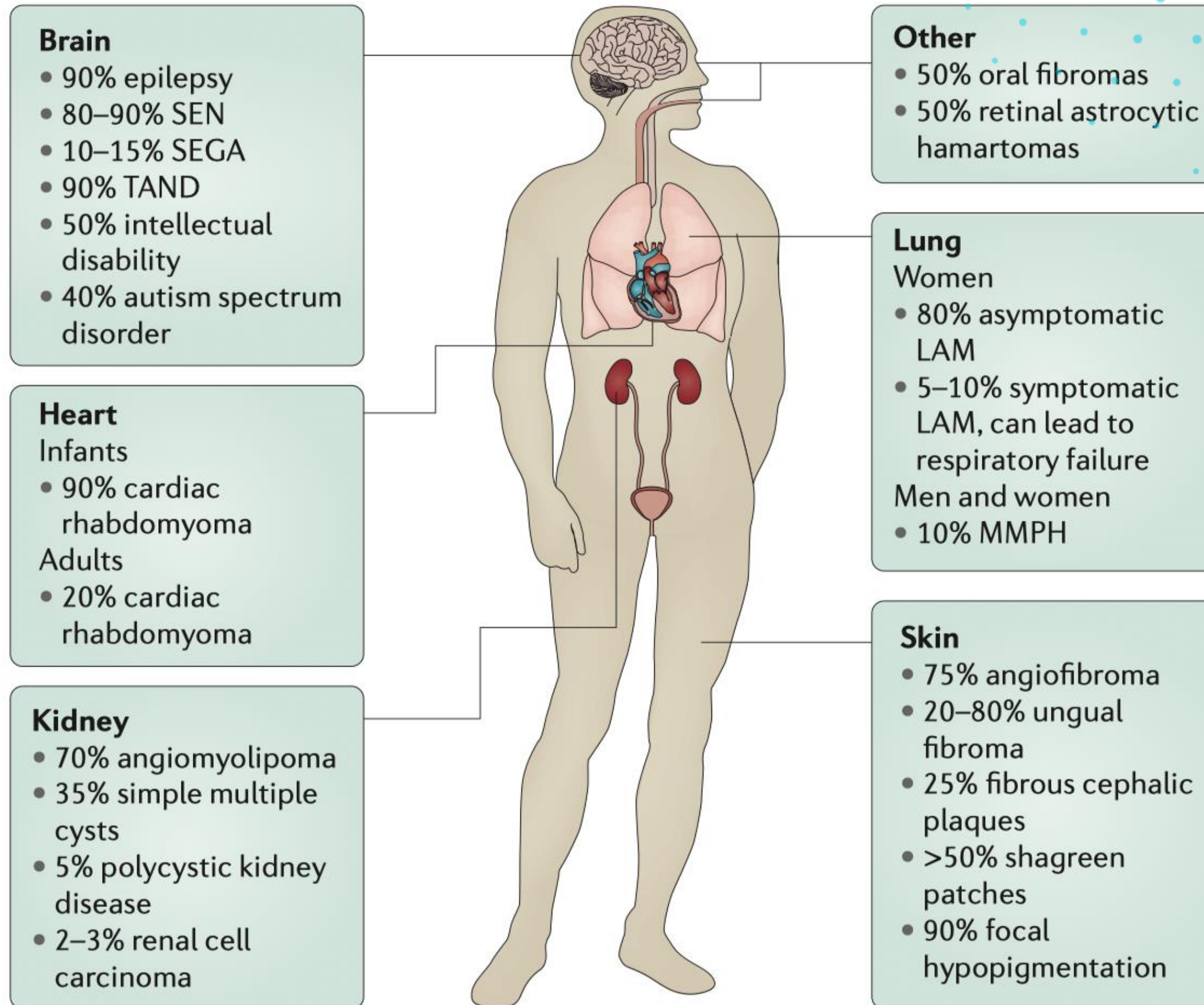
Placa fibrosa cefálica

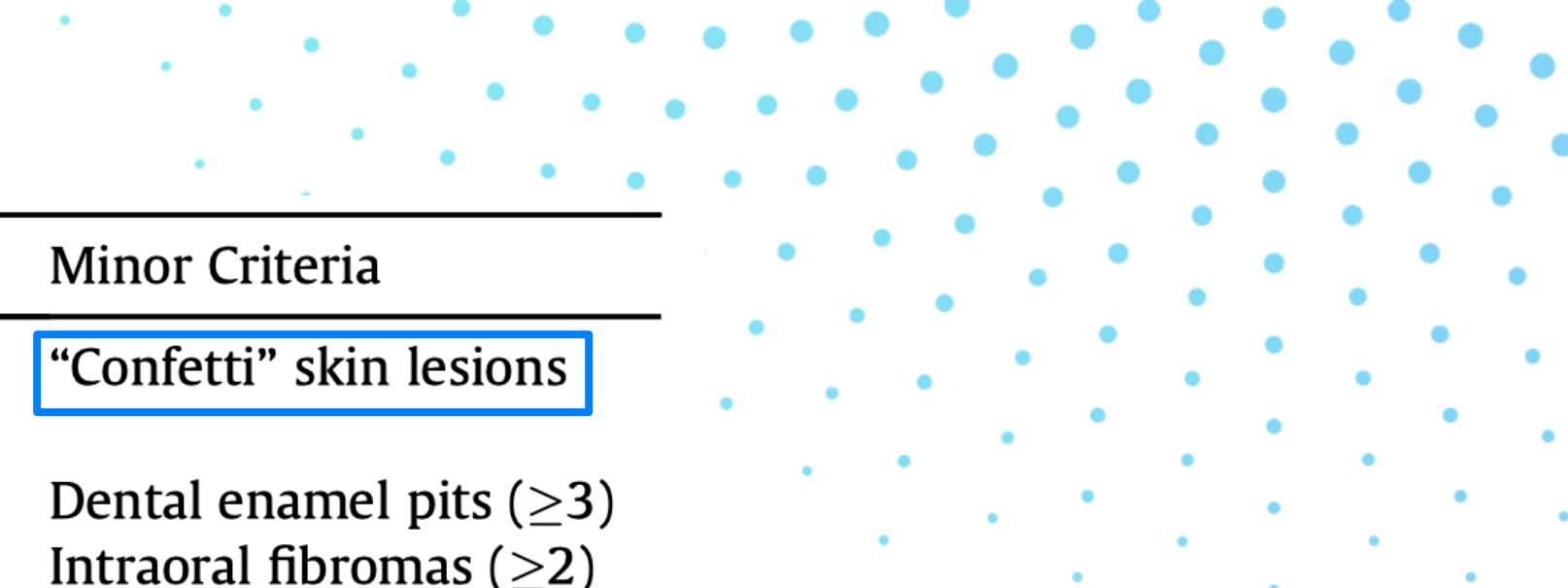


Angiofibromas faciais



Fibromas gengivais





Major Criteria	Minor Criteria
Hypomelanotic macules (≥ 3; at least 5 mm diameter) Angiofibroma (≥ 3) or fibrous cephalic plaque Ungual fibromas (≥ 2) Shagreen patch Multiple retinal hamartomas Multiple cortical tubers and/or radial migration lines Subependymal nodule (≥ 2) Subependymal giant cell astrocytoma Cardiac rhabdomyoma LAM* Angiomyolipomas (≥ 2)*	“Confetti” skin lesions Dental enamel pits (≥ 3) Intraoral fibromas (≥ 2) Retinal achromic patch Multiple renal cysts Nonrenal hamartomas Sclerotic bone lesions

Abbreviations:

LAM = Lymphangiomyomatosis

TSC = Tuberous sclerosis complex

Definite TSC: 2 major features or 1 major feature with 2 minor features.

Possible TSC: either 1 major feature or ≥ 2 minor features.

Genetic diagnosis: A pathogenic variant in *TSC1* or *TSC2* is diagnostic for TSC (most TSC-causing variants are sequence variants that clearly prevent TSC1 or TSC2 protein production. Some variants compatible with protein production [e.g., some missense changes] are well established as disease-causing; other variant types should be considered with caution).

Tratamento



Acompanhamento multidisciplinar:

Neurologista
Cardiologista
Oftalmologista
Pneumologista
Nefrologista



Aconselhamento genético

Tratamento



Manifestações cutâneas

Eletrocoagulação

Excisão/Shaving

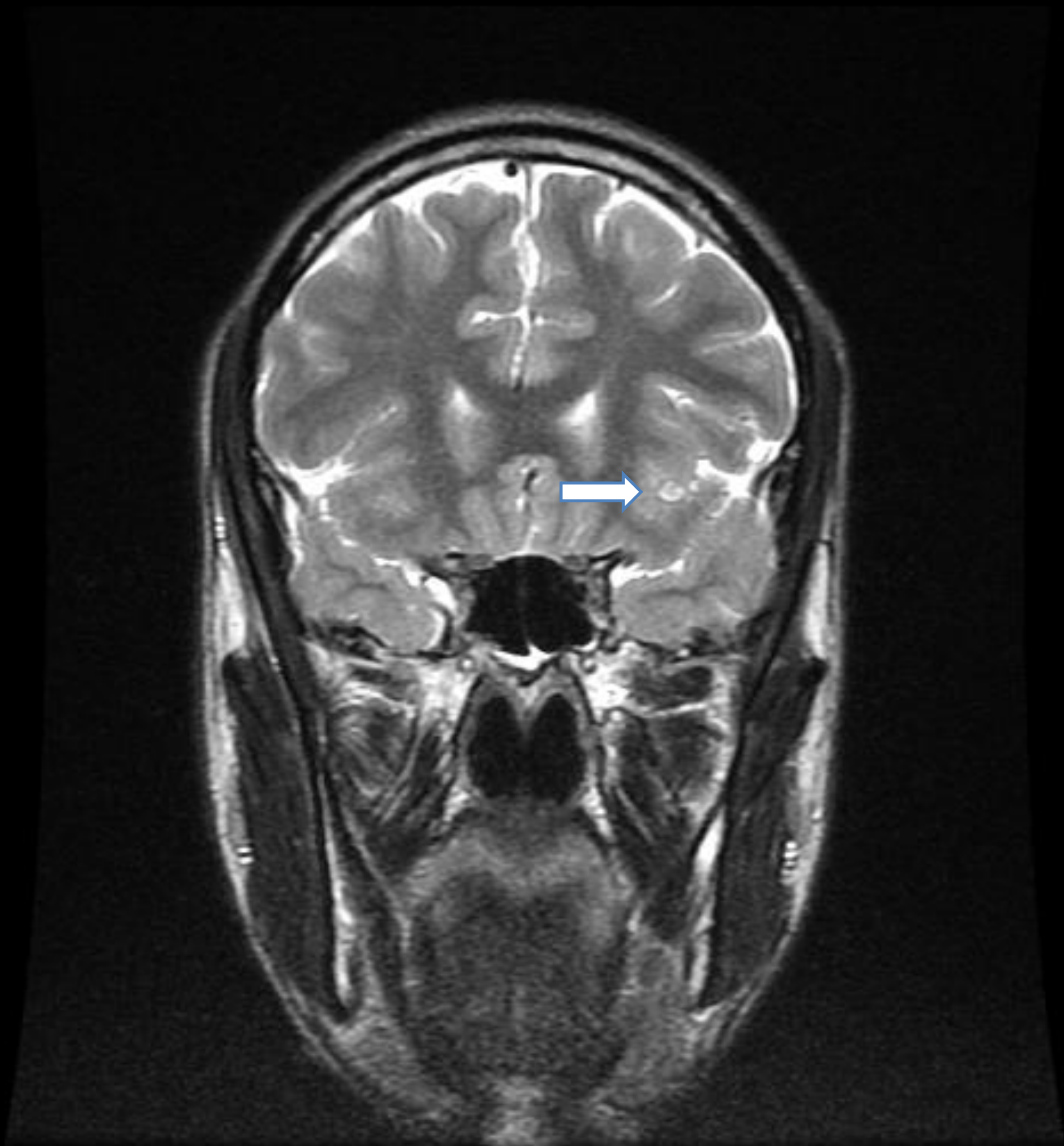
Crioterapia

Dermabrasão

Laser



Everolimus e Sirolimus: angiomiolipoma renal, linfangioleiomiomatose pulmonar e astrocitoma
Terapia tópica com Sirolimus em gel: aprovada pelo FDA para angiofibromas faciais a partir dos 6 anos



**Mas e a
Doença de Crohn?**

Dois artigos na literatura relataram associação entre as duas doenças

A rare association of tuberous sclerosis and Crohn's disease:
Report of two cases

Narendra Vadlamudi, Martina Bebin, Kirk Thame, Jose Mestre
Children's of Alabama, Birmingham, USA

Case report



J Korean Child Neurol Soc 2018;26(4):284-287
<https://doi.org/10.26815/jkcns.2018.26.4.284>
pISSN 1226-6884 • eISSN 2383-8973

Tuberous Sclerosis Complex with Crohn's Disease

Tuberous sclerosis complex (TSC) is an autosomal dominant disorder caused by

Min Hee Kim¹, Yoon Jin Lee, MD², Jae Young Kim,
MD, PhD³, Yoon Young Yi, MD², Joon Won Kang,
MD, PhD^{2,4}

Original Article

**Tuberous Sclerosis Complex 1 Deficiency in Macrophages
Promotes Unclassical Inflammatory Response to
Lipopolysaccharide *In Vitro* and Dextran Sodium Sulfate-
Induced Colitis in Mice**

**Huawen Xu^{1,2,#}, Yang Zhao^{1,2,#}, Qingjie Zhao^{1,3,#}, Mingpu Shi^{1,2,#}, Zhaoqi Zhang^{1,2}, Wenjun
Ding^{3,*}, Yong Zhao^{1,2,4,*}**

Motivos da apresentação



Destacar o papel do dermatologista no diagnóstico da esclerose tuberosa

Ressaltar a importância de ir além da queixa principal do paciente e realizar um exame físico detalhado

Demonstrar uma possível relação entre esclerose tuberosa e Doença de Crohn

Referências

1. Curatolo P, Bombardieri R, Jozwiak S. Tuberous Sclerosis. The Lancet. 2008 Aug;372(9639):657–68.
2. Xu H, Zhao Y, Zhao Q, Shi M, Zhang Z, Ding W, et al. Tuberous Sclerosis Complex 1 Deficiency in Macrophages Promotes Unclassical Inflammatory Response to Lipopolysaccharide In Vitro and Dextran Sodium Sulfate-Induced Colitis in Mice. Aging and Disease. 2022 Jan 1;13(6):1875–5.
3. Stuart C, Fladrowski C, Flinn J, Öberg B, Peron A, Rozenberg M, et al. Beyond the Guidelines: How We Can Improve Healthcare for People With Tuberous Sclerosis Complex Around the World. Pediatric Neurology. 2021 Oct;123(123):77–84.
4. Portocarrero LKL, Quental KN, Samorano LP, de Oliveira ZNP, Rivitti-Machado MC da M. Tuberous sclerosis complex: review based on new diagnostic criteria. Anais Brasileiros de Dermatologia. 2018;93(3):323–31.
5. Henske EP, Jóźwiak S, Kingswood JC, Sampson JR, Thiele EA. Tuberous sclerosis complex. Nature Reviews Disease Primers. 2016 May 26;2(1).
6. Vadlamudi N, Bebin M, Thame K, Mestre J. A rare association of tuberous sclerosis and Crohn's disease: Report of two cases. Inflammatory Bowel Diseases. 2011 Dec;17(2):S62–3.
7. Kim MH, Lee YJ, Kim JY, Yi YY, Kang JW. Tuberous Sclerosis Complex with Crohn's Disease. Child Neurology. 2018 Dec 31;26(4):284–7.

Obrigado!