

「TSC」

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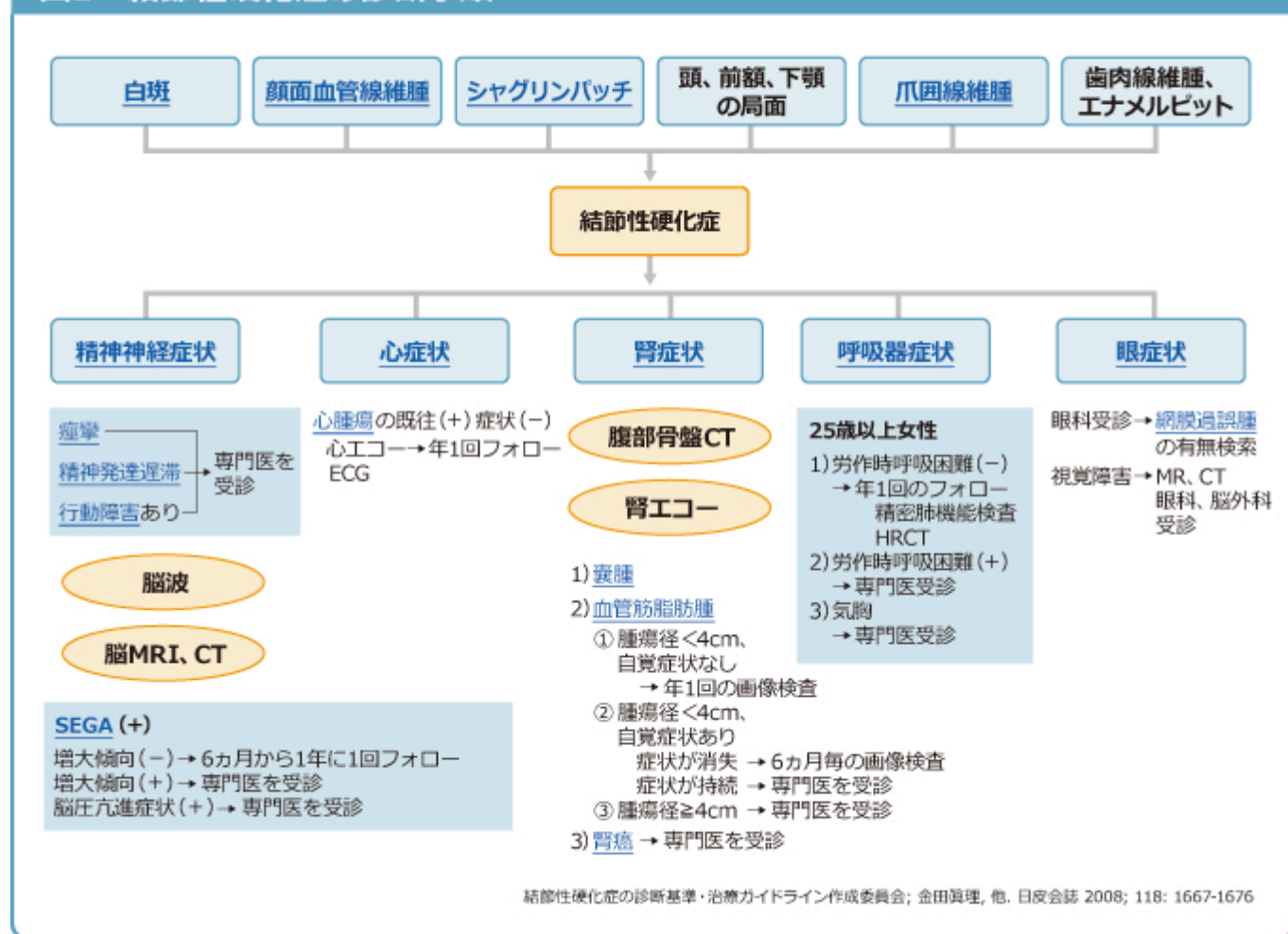
「TSC」

「TSC」1835「TSC」1935「TSC」50「TSC」1993「TSC2」1997「TSC1」2000「TSC1」「TSC2」PI3K-Akt-mTOR「TSC」2001「TSC」2002「TSC」2008¹⁾「TSC」1998「TSC」1「TSC Clinical Consensus Conference」Roach(「Gometz」)

「TSC」1

Image

図1 結節性硬化症の診断手順



2012「TSC」2「TSC」Consensus Conference「TSC」1「TSC」2²⁾³⁾「TSC」mTOR「TSC」2

Consensus Conference 2018 National Comprehensive Cancer Network (NCCN) Clinical guideline 2018 4) 1

1 4

A

TSC1/TSC2 TSC

B

1 3 5mm

2 3

3 2 ungual fibromas

4 shagreen patch/connective tissue nevus

5 multiple retinal nodular hamartomas

6 cortical tube cerebral white matter radial migration lines

7 subependymal nodule

8 subependymal giant cell astrocytoma

9 cardiac rhabdomyoma

10 lymphangioleiomyomatosis LAM⁰¹

11 angiomyolipoma 2⁰¹

1 confetti skin lesions

2 3 multiple randomly distributed dental enamel pits

3 2 intraoral fibromas

4 retinal achromic patch

5 multiple renal cyst

6 nonrenal hamartoma

1 lymphangioleiomyomatosis renal angiomyolipoma Definitive TSC

Definitive TSC 2 1 2

Possible TSC 1 2

1) 2008; 118: 1667-1676

2) Northrup H, et al. Pediatr Neurol 2013; 49: 243-254

3) Krueger DA, et al. N Engl J Med 2013; 48: 255-265

4) 2018; 128: 1-16

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