

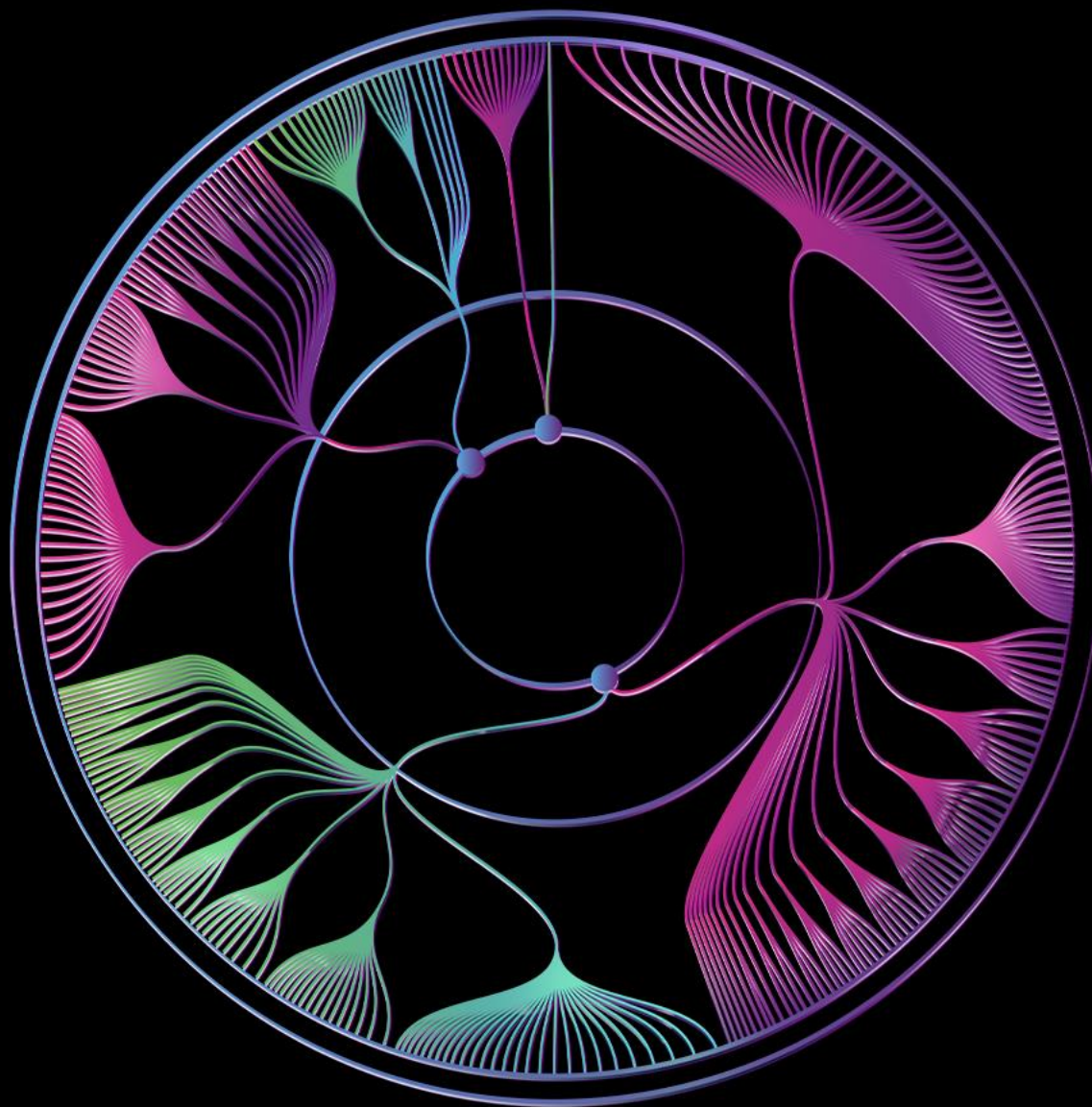
CBR22 51º CONGRESSO BRASILEIRO
DE RADIOLOGIA E
DIAGNÓSTICO POR IMAGEM

CASO Nº. 09

Patrocínio
educacional:



Colégio Brasileiro de Radiologia
e Diagnóstico por Imagem



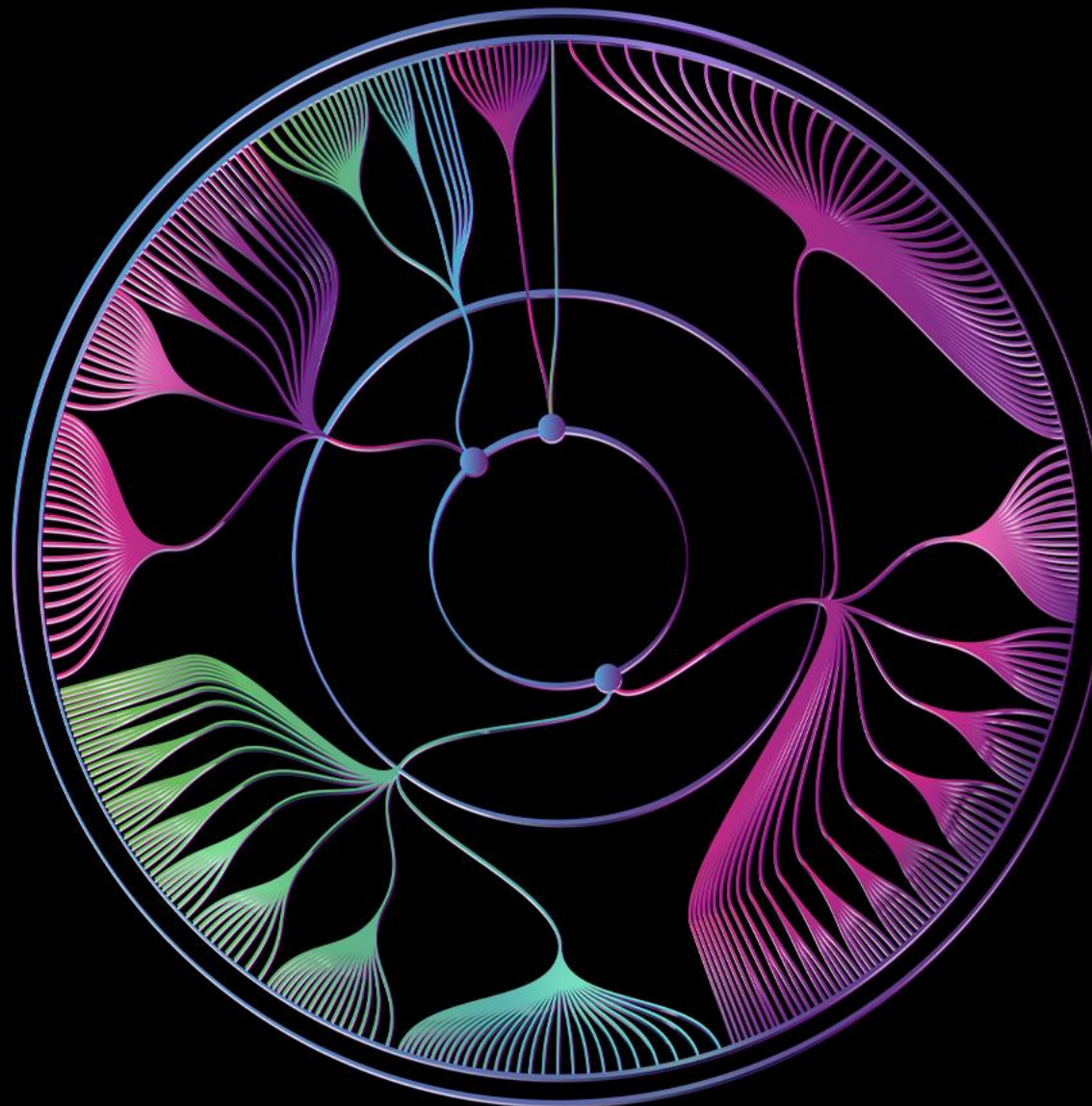
CBR22

51º CONGRESSO BRASILEIRO
DE RADIOLOGIA E
DIAGNÓSTICO POR IMAGEM

Subespecialidade:

PEDIATRIA

**Caso gentilmente cedido pela Dra.
Maria Clara Zanon Zotin**



cbr

Colégio Brasileiro de Radiologia
e Diagnóstico por Imagem

RN, sexo feminino, natural de Ribeirão Preto - SP.

Ao nascimento:

Idade gestacional 36 semanas e 5 dias. Pré-natal irregular, com poucas informações.

1870g, 43 cm, PC 35 cm

APGAR: 9/9

Parto cesáreo por sofrimento fetal + iteratividade. FC >100 BPM. Encaminhada ao CTI neonatal.

Ao exame: Bom estado geral, corada, hidratada, acianótica, ativa e reativa.

FR 66, FC 132, glicemia capilar 19, temperatura axilar 36,4 graus. Pressão arterial: MSD (57x20), MID (46x25), MSE (45x28), MIE (48x22).

Ap. Respiratório: retração subcostal discreta.

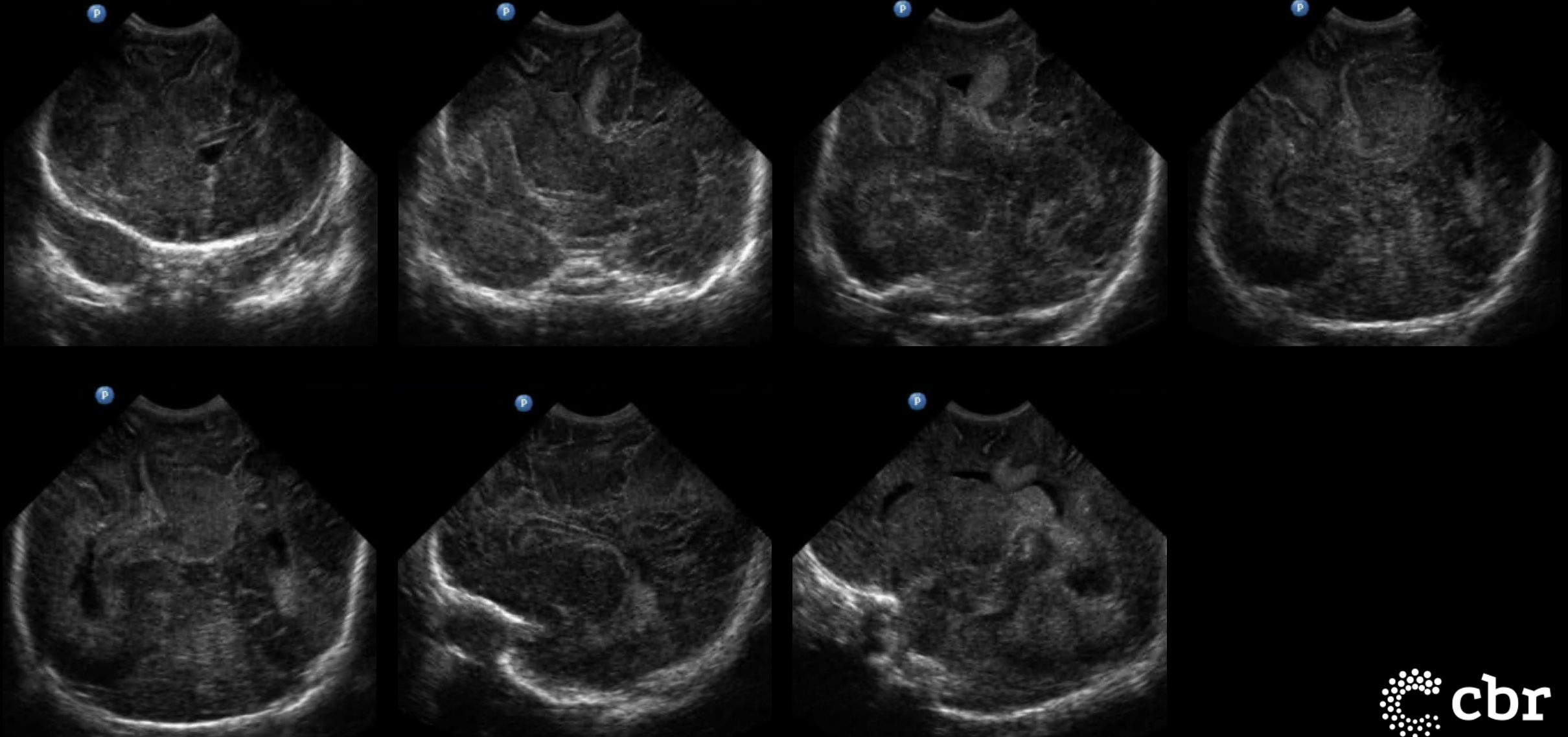
Ap. Cardiovascular: sopro sistólico audível em todos os focos, principalmente aórtico.

Abdome: nada digno de nota.

Abaulamento parietal e discreta exoftalmia à direita.

Genitália típica feminina, ânus tópico e pérvio.

US Transfontanelar

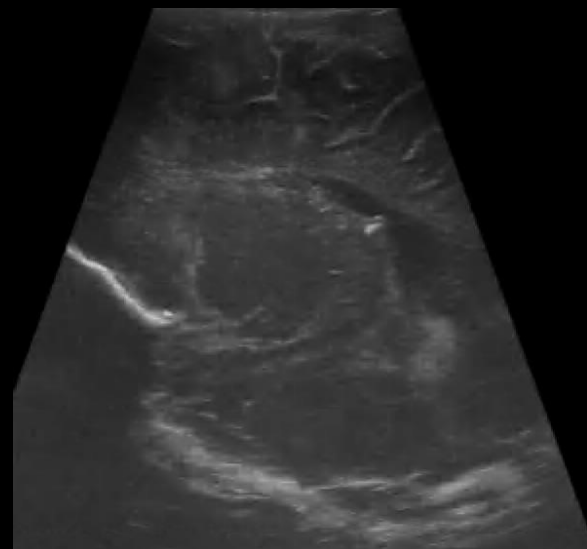
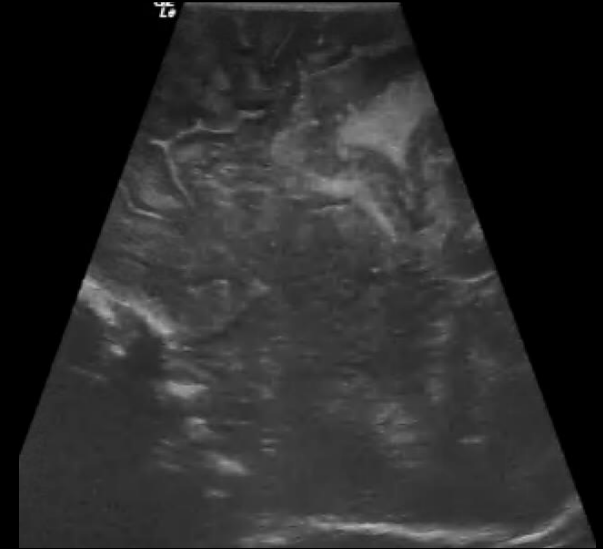
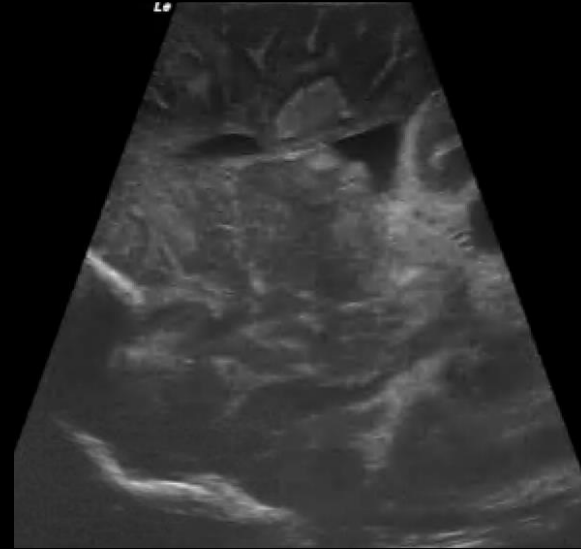
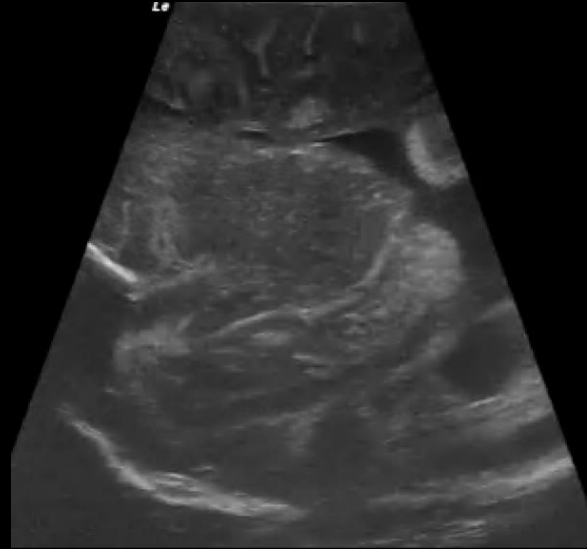


US Transfontanelar

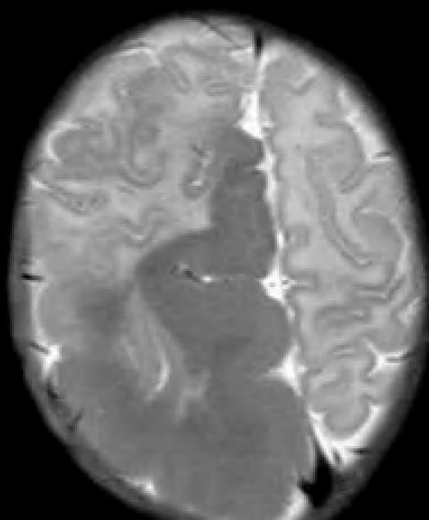


US Transfontanelar

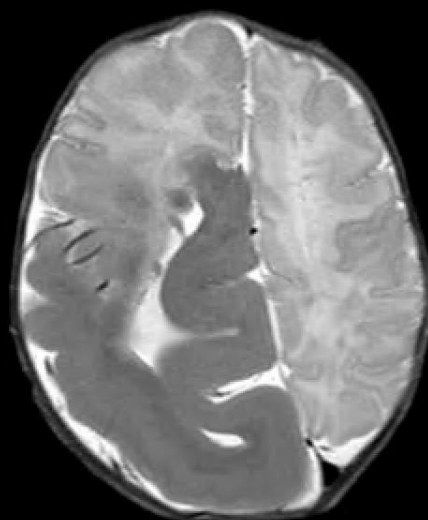
CBR22 51º CONGRESSO BRASILEIRO
DE RADIOLOGIA E
DIAGNÓSTICO POR IMAGEM



Ressonância Magnética (com 21 dias)



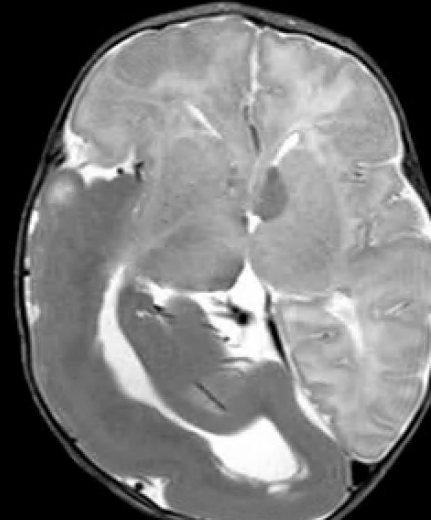
Axial T2



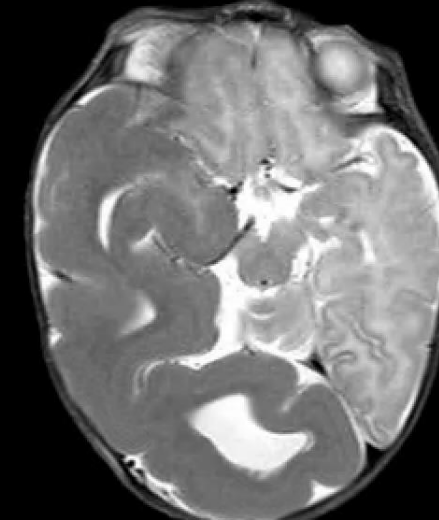
Axial T2



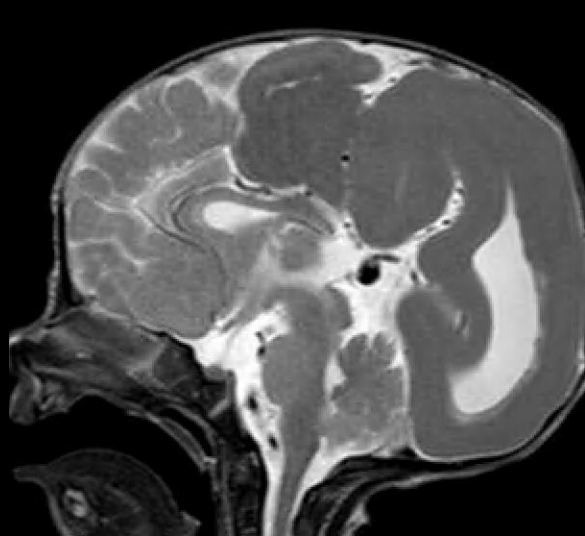
Axial T2



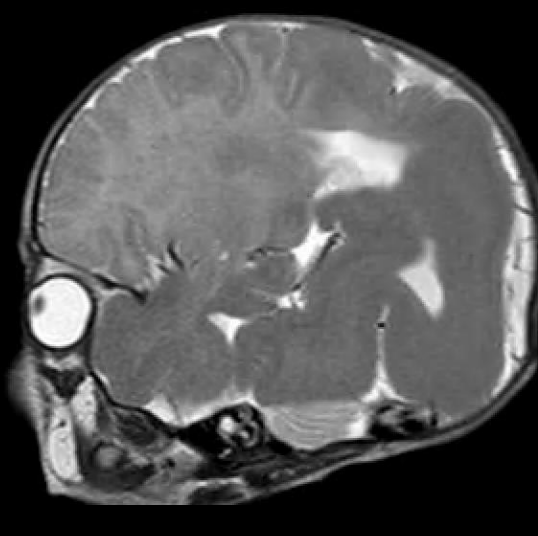
Axial T2



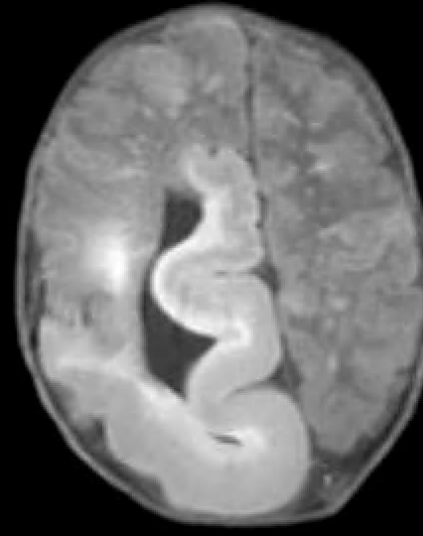
Axial T2



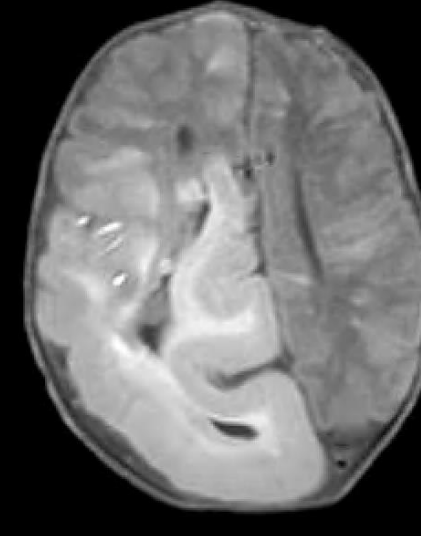
Sagittal T2



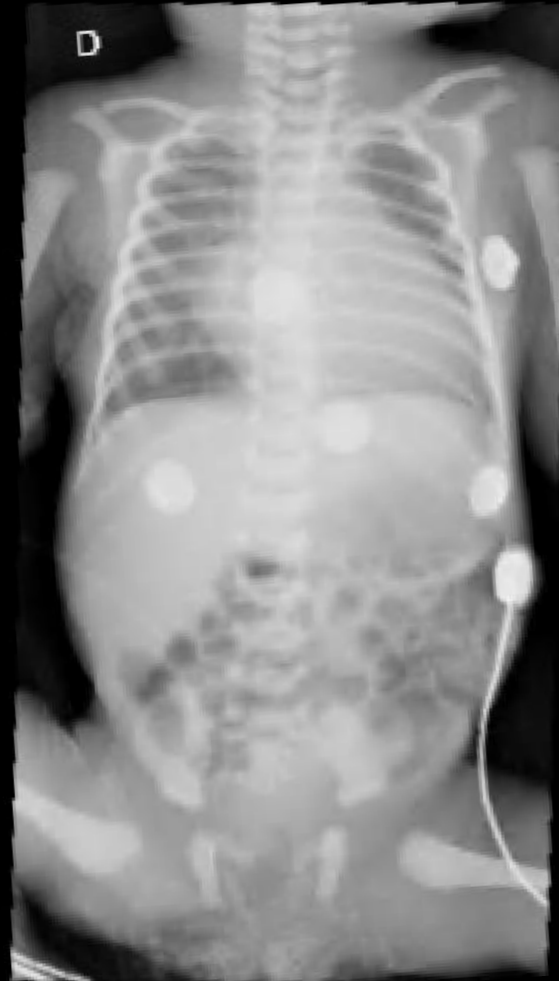
Sagittal T2

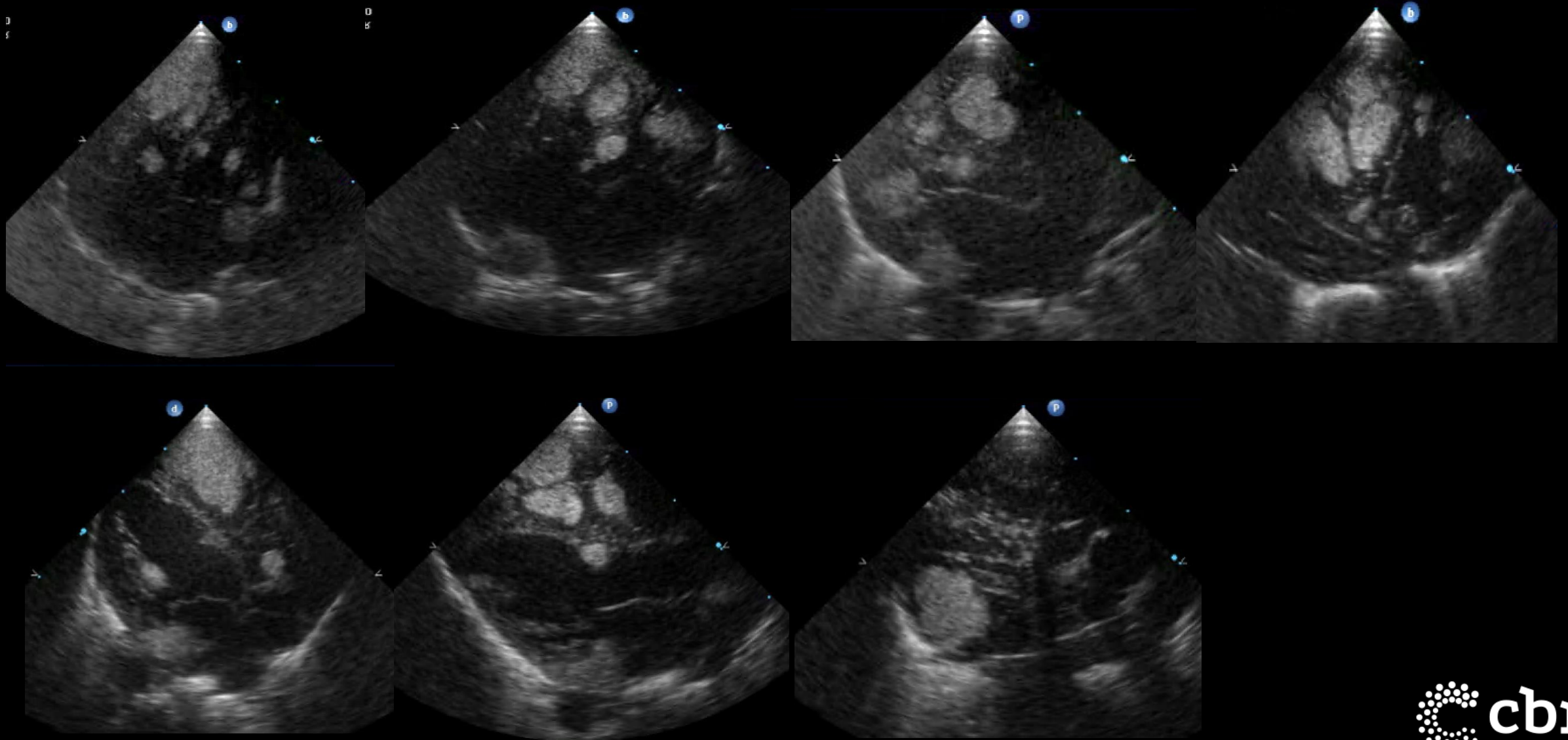


Axial T1

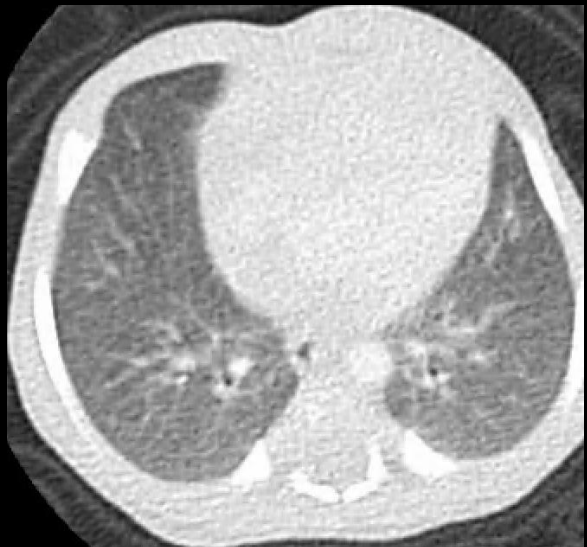
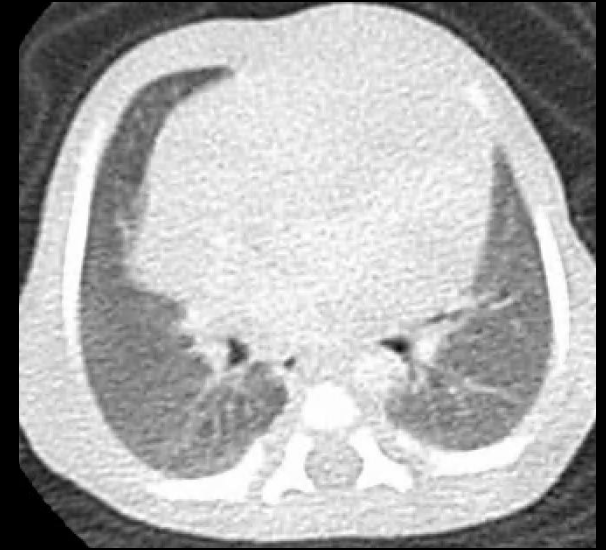


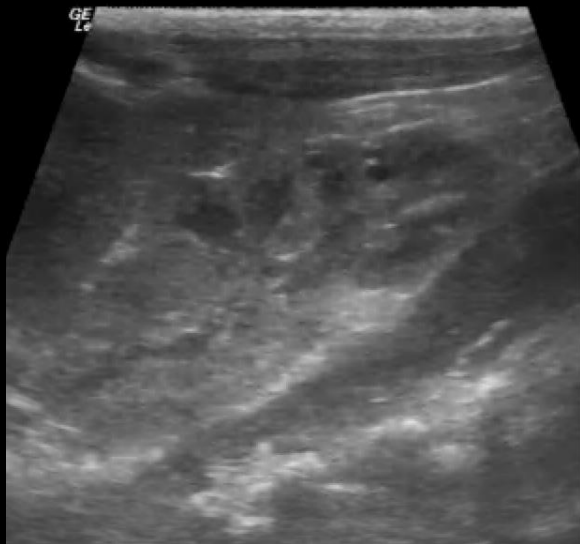
Axial T1



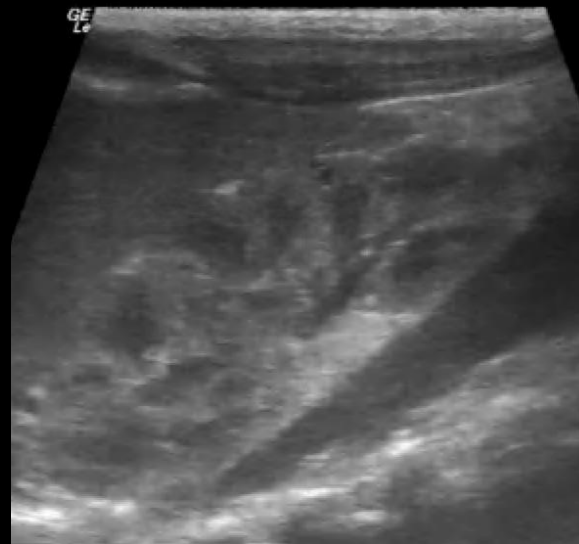


Tomografia de tórax

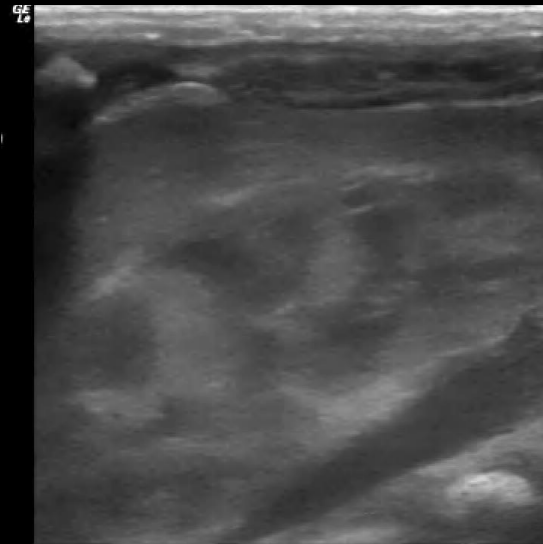




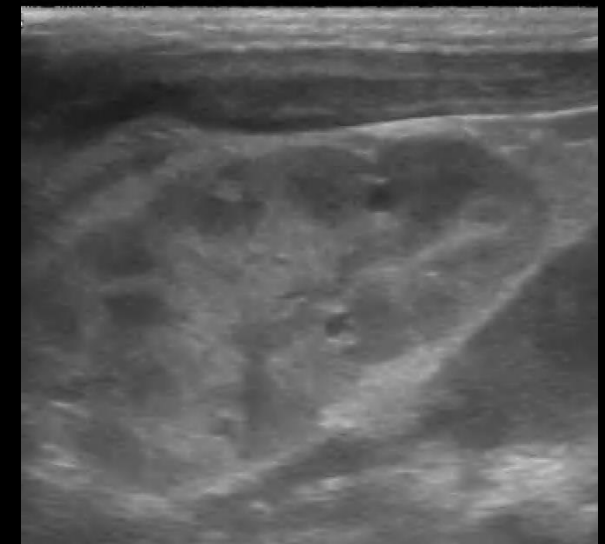
Rim direito



Rim direito



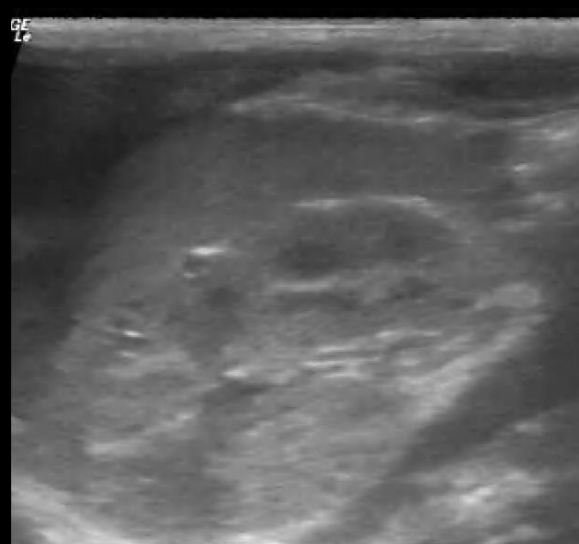
Rim direito



Rim direito



Rim esquerdo



Rim esquerdo

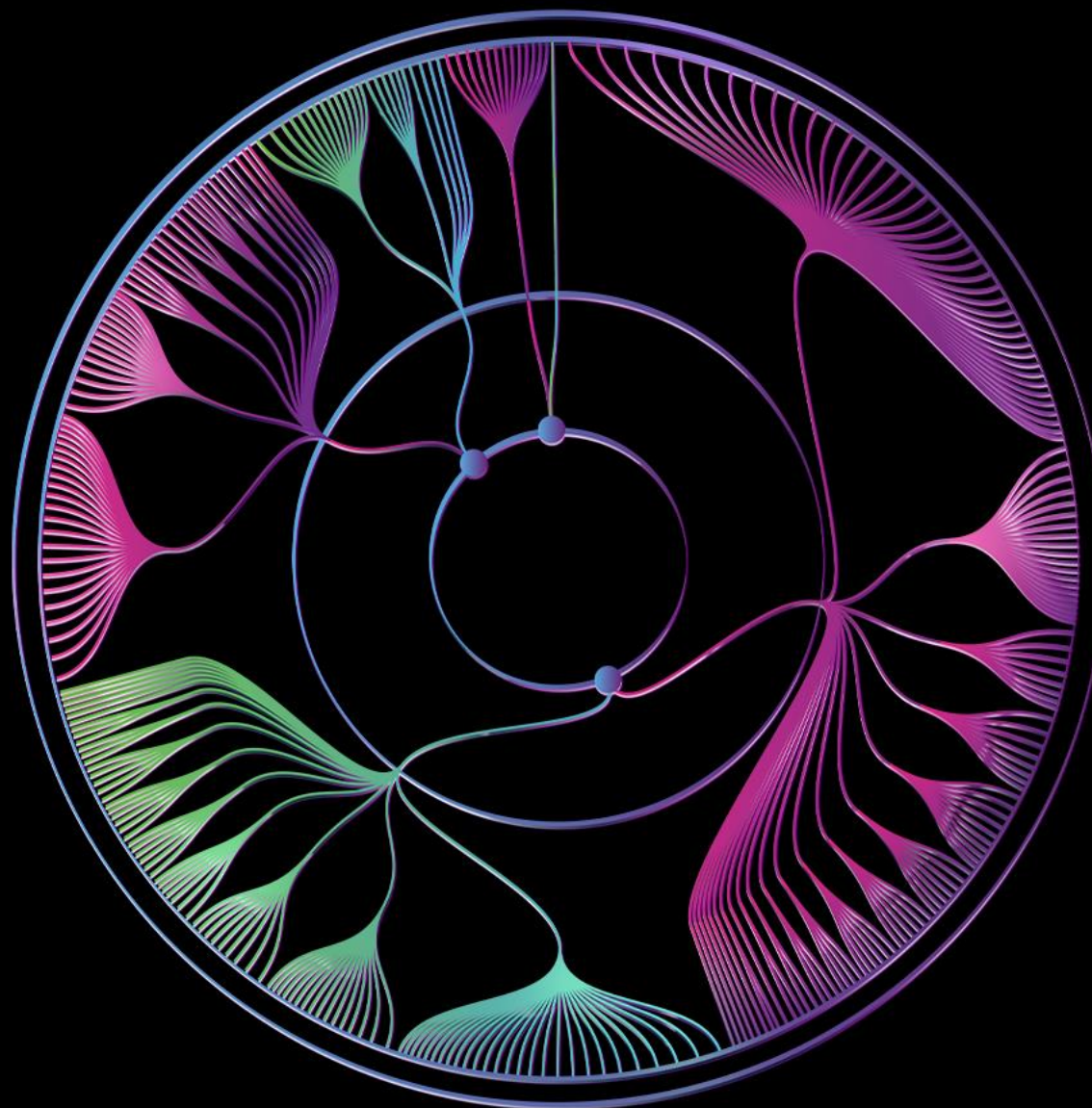
Qual o seu diagnóstico?

- a) Neurofibromatose**
- b) Síndrome de von Hippel Lindau**
- c) Síndrome de Sturge Weber**
- d) Esclerose Tuberosa**
- e) Síndrome de Lowe**

CBR22

51º CONGRESSO BRASILEIRO
DE RADIOLOGIA E
DIAGNÓSTICO POR IMAGEM

**A partir deste slide só será
publicado após o fim da
Maratona**

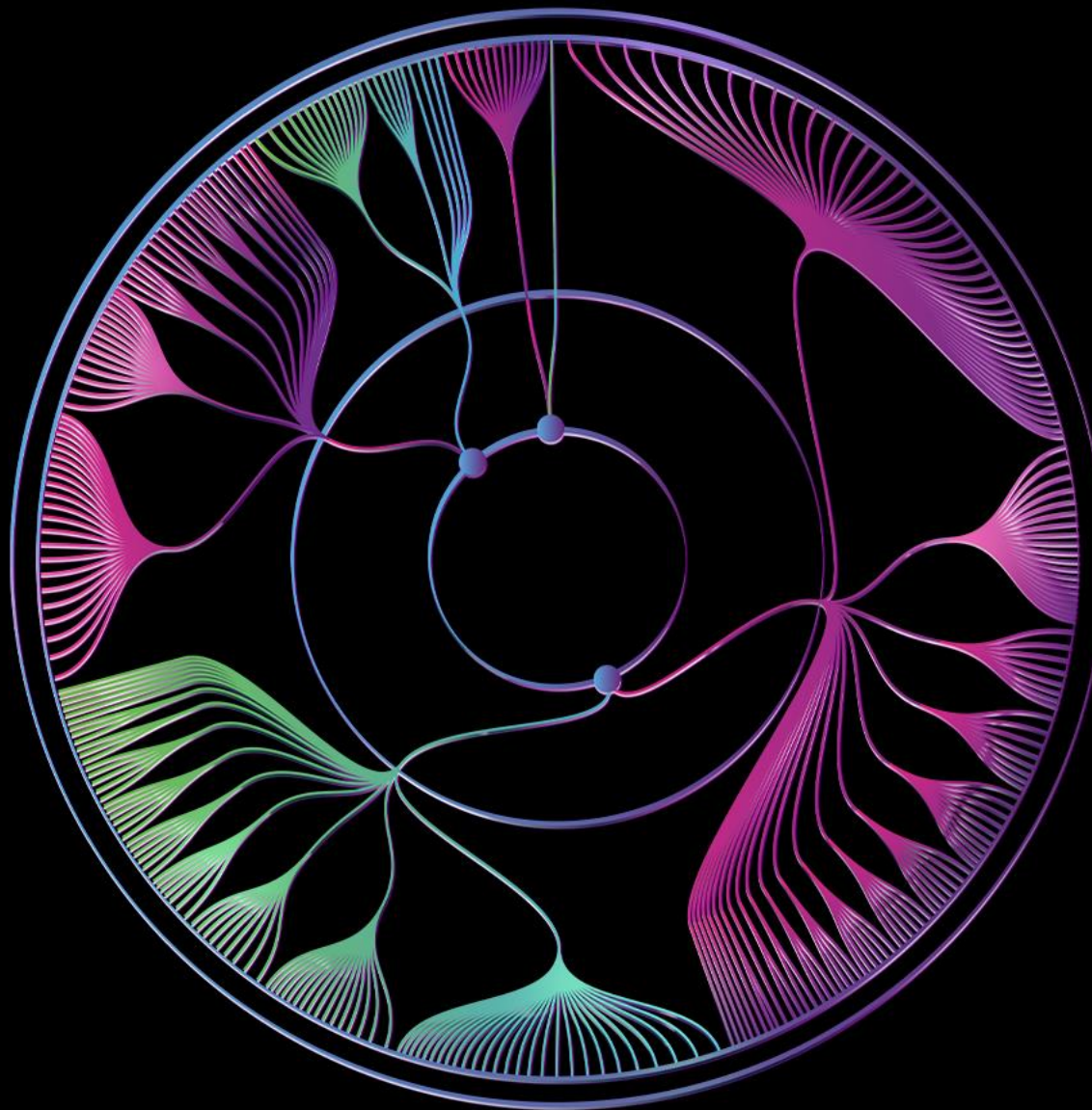


CBR22

51º CONGRESSO BRASILEIRO
DE RADIOLOGIA E
DIAGNÓSTICO POR IMAGEM

Gabarito

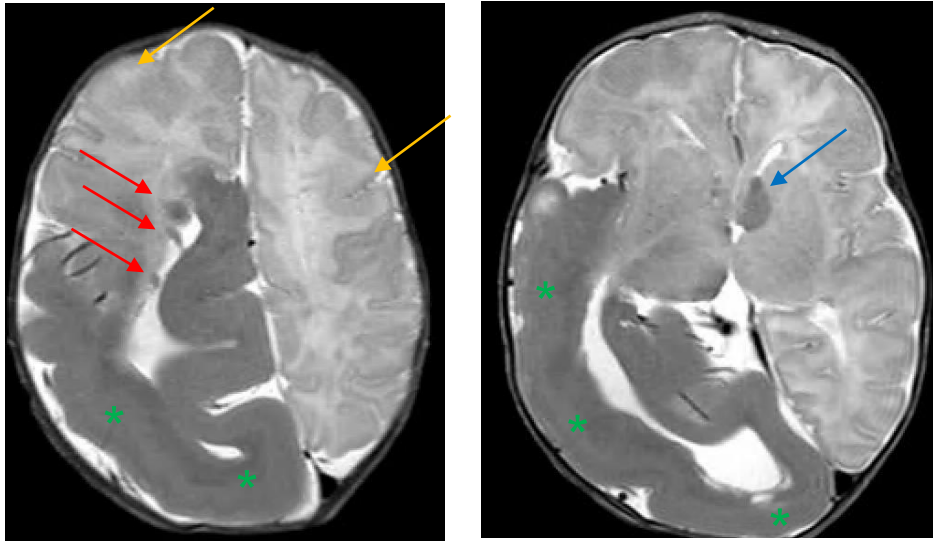
**Subespecialidade:
Pediatria**








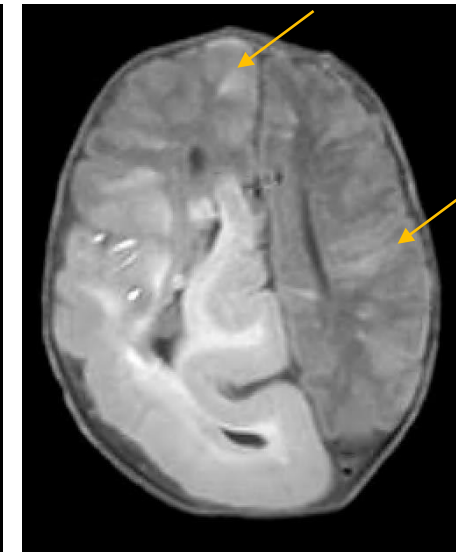
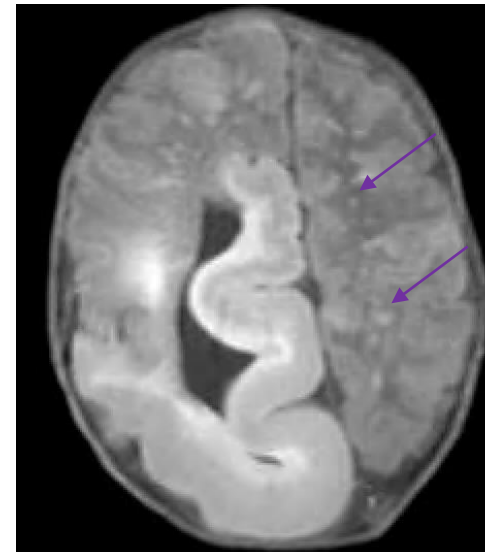
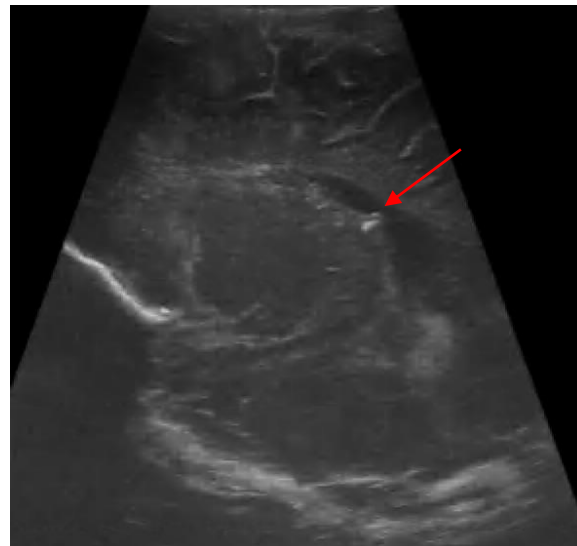
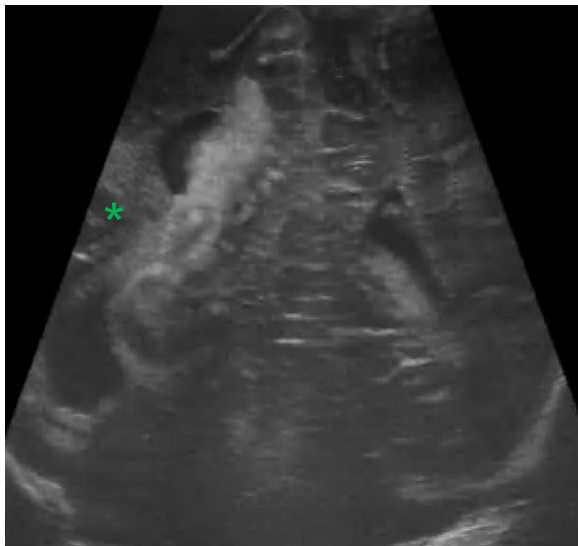
cbr

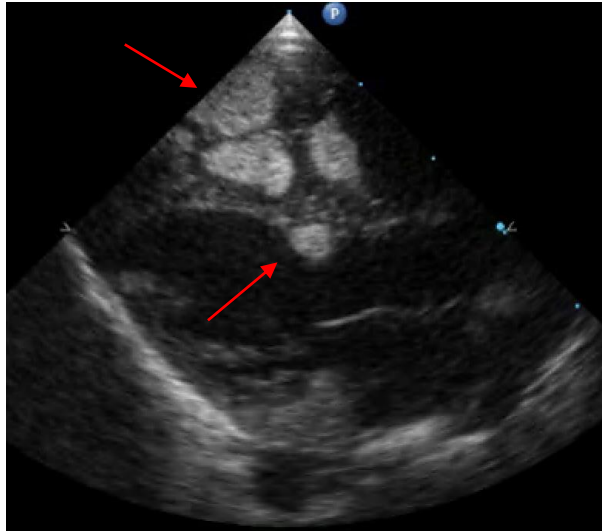
Colégio Brasileiro de Radiologia
e Diagnóstico por Imagem



- a) Neurofibromatose
- b) Síndrome de von Hippel Lindau
- c) Síndrome de Sturge Weber
- d) Esclerose Tuberosa**
- e) Síndrome de Lowe

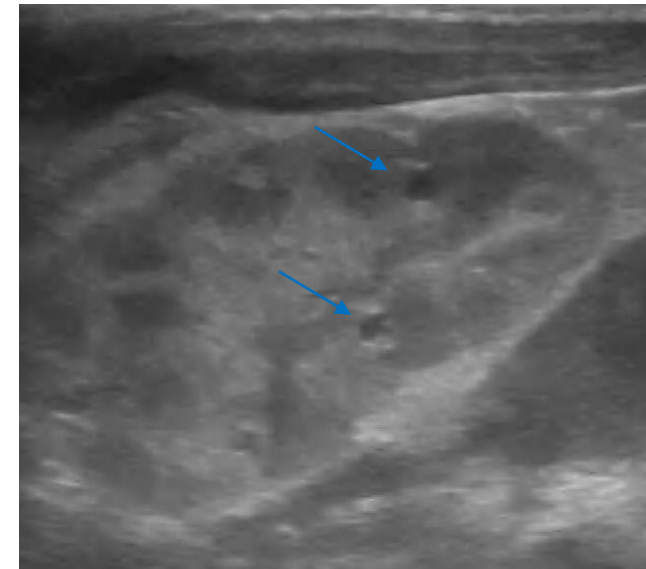
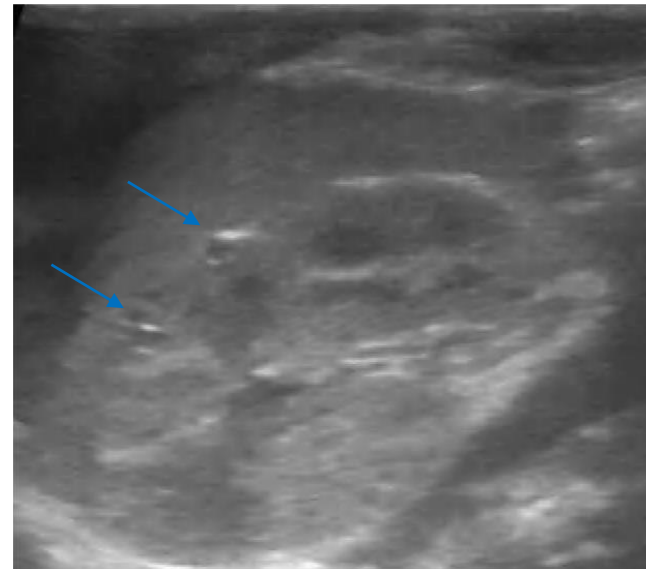
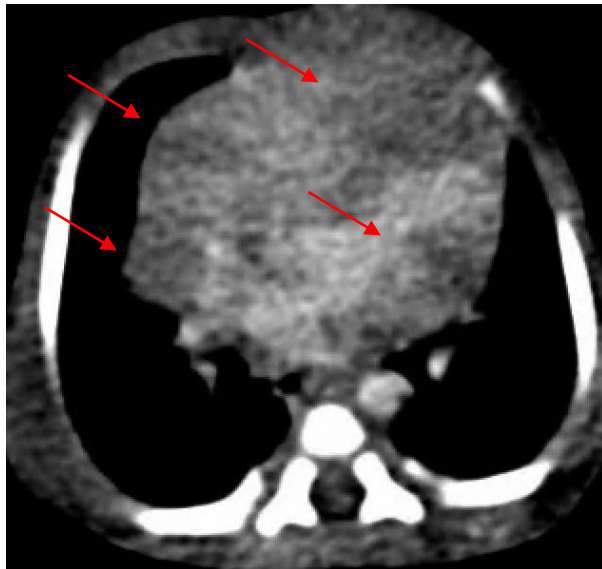


-  Nódulos subependimários
-  Tumor subependimário de células gigantes
-  Áreas de displasia cortical
-  Anomalias da substância branca
-  Hemimegalencefalia





-  **Rabdomiomas cardíacos**
-  **Cistos renais**



Esclerose Tuberosa:

Síndrome neurocutânea autossômica dominante relativamente rara, causada por mutação nos genes supressores tumorais TSC1 ou TSC2.

Manifestações mais comuns:

- **Túberes corticais e subcorticais**
- Heterotopia de substância cinzenta
- **Nódulos subependimários**
- **Tumor subependimário de células gigantes**
- **Rabdomiomas cardíacos**
- Linfangioleiomiomatose
- Angiomiolipomas renais
- **Cistos renais**
- Carcinoma renal

E a hemimegalencefalia?

NeuroImages

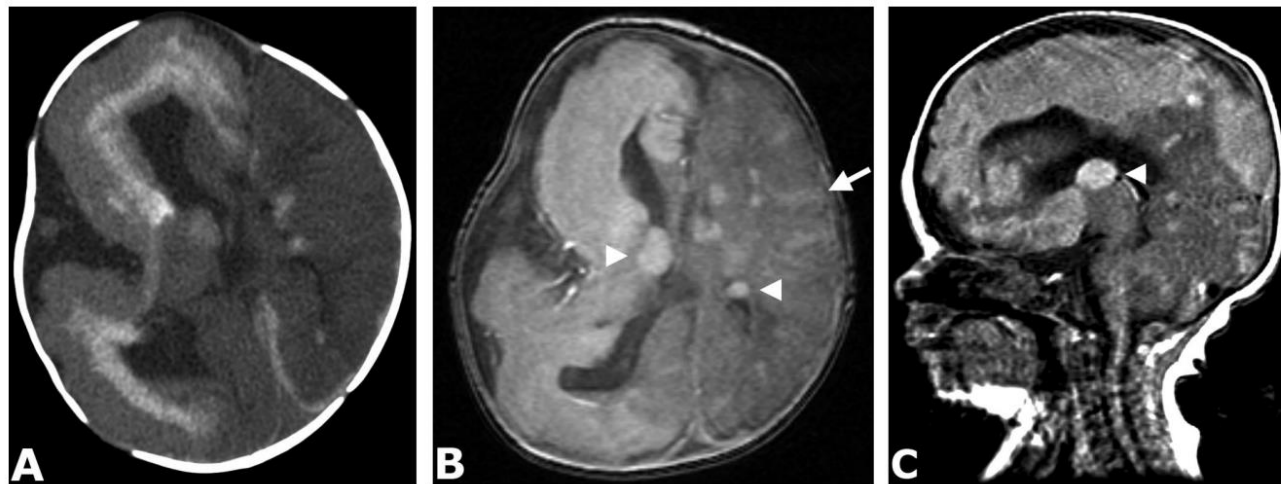


Figure. CT without contrast (A) shows hemimegalencephaly and calcified heterotopia outlining the right lateral ventricle. Axial T1-weighted MRI with contrast (B) and sagittal T1-weighted MRI without contrast (C) demonstrate cortical dysplasia in the left hemisphere (arrow) and subependymal nodules (arrowheads).

Hemimegalencephaly and tuberous sclerosis complex

Michael S. Cartwright, MD; Sean C. McCarthy, BA; and E. Steve Roach, MD, Winston-Salem, NC

A 1-day-old boy was transferred to the neonatal intensive care unit secondary to tachypnea. He had macrocephaly and full fontanelles. Imaging demonstrated hemimegalencephaly and subependymal nodules (figure). Further evaluation revealed cardiac rhabdomyomas and renal angiomyolipomas. Definite tuberous

sclerosis complex was diagnosed on the basis of the presence of two major features.¹

Hemimegalencephaly and tuberous sclerosis complex are distinct and rare conditions that are characterized by cortical malformations. While they are currently thought to be unrelated, there are similar cases in the literature² and it is conceivable that an abnormality early in cortical development could lead to both conditions in an individual.

Copyright © 2005 by AAN Enterprises, Inc.

1. Roach ES, Gomez MR, Northrup H. Tuberous sclerosis complex consensus conference: revised clinical diagnostic criteria. *J Child Neurol* 1998; 13:624–628.
2. Galluzzi P, Cerase A, Strambi M, Buoni S, Fois A, Venturi C. Hemimegalencephaly in tuberous sclerosis complex. *J Child Neurol* 2002;17:677–680.

Address correspondence and reprint requests to Dr. Michael S. Cartwright, Wake Forest University School of Medicine, Department of Neurology, Medical Center Blvd., Winston-Salem, NC 27157; e-mail: mcartwri@wfubmc.edu

Hemimegalencephaly and tuberous sclerosis complex

Michael S. Cartwright, Sean C. McCarthy, E. Steve Roach. *Neurology* May 2005, 64 (9) 1634.

Galluzzi P, Cerase A, Strambi M, Buoni S, Fois A, Venturi C. Hemimegalencephaly in tuberous sclerosis complex. *J Child Neurol*. 2002 Sep;17(9):677-80. doi: 10.1177/088307380201700905. PMID: 12503644.

Sidira C, Vargiami E, Dragoumi P, Zafeiriou DI. Hemimegalencephaly and tuberous sclerosis complex: A rare yet challenging association. *Eur J Paediatr Neurol*. 2021 Jan;30:58-65. doi: 10.1016/j.ejpn.2020.12.007. Epub 2020 Dec 23. PMID: 33387903.

Parmar H, Patkar D, Shah J, Patankar T. Hemimegalencephaly with tuberous sclerosis: a longitudinal imaging study. *Australas Radiol*. 2003 Dec;47(4):438-42. doi: 10.1046/j.1440-1673.2003.01216.x. PMID: 14641199.

A associação entre esclerose tuberosa e hemimegalencefalia é incomum, mas já foi relatada em alguns casos.

1. Wang, Segaran, N., Bhalla, S., Pickhardt, P. J., Lubner, M. G., Katabathina, V. S., & Ganeshan, D. (2021). Tuberos Sclerosis: Current Update. *Radiographics*, 41(7), 1992–2010.
<https://doi.org/10.1148/rq.2021210103>
2. Baron Y, Barkovich AJ. MR imaging of tuberous sclerosis in neonates and young infants. *AJNR Am J Neuroradiol*. 1999 May;20(5):907-16. PMID: 10369365; PMCID: PMC7056154.