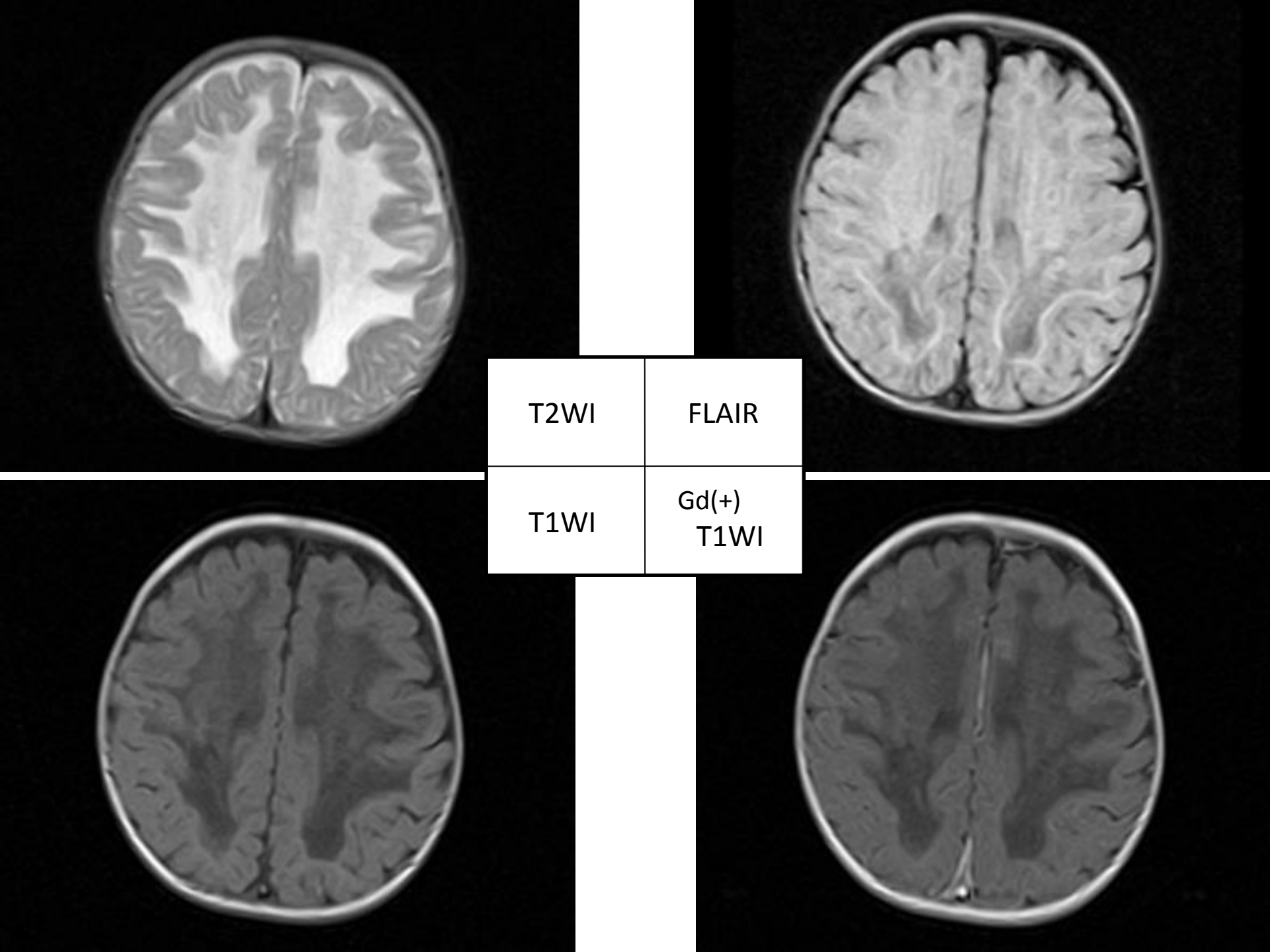
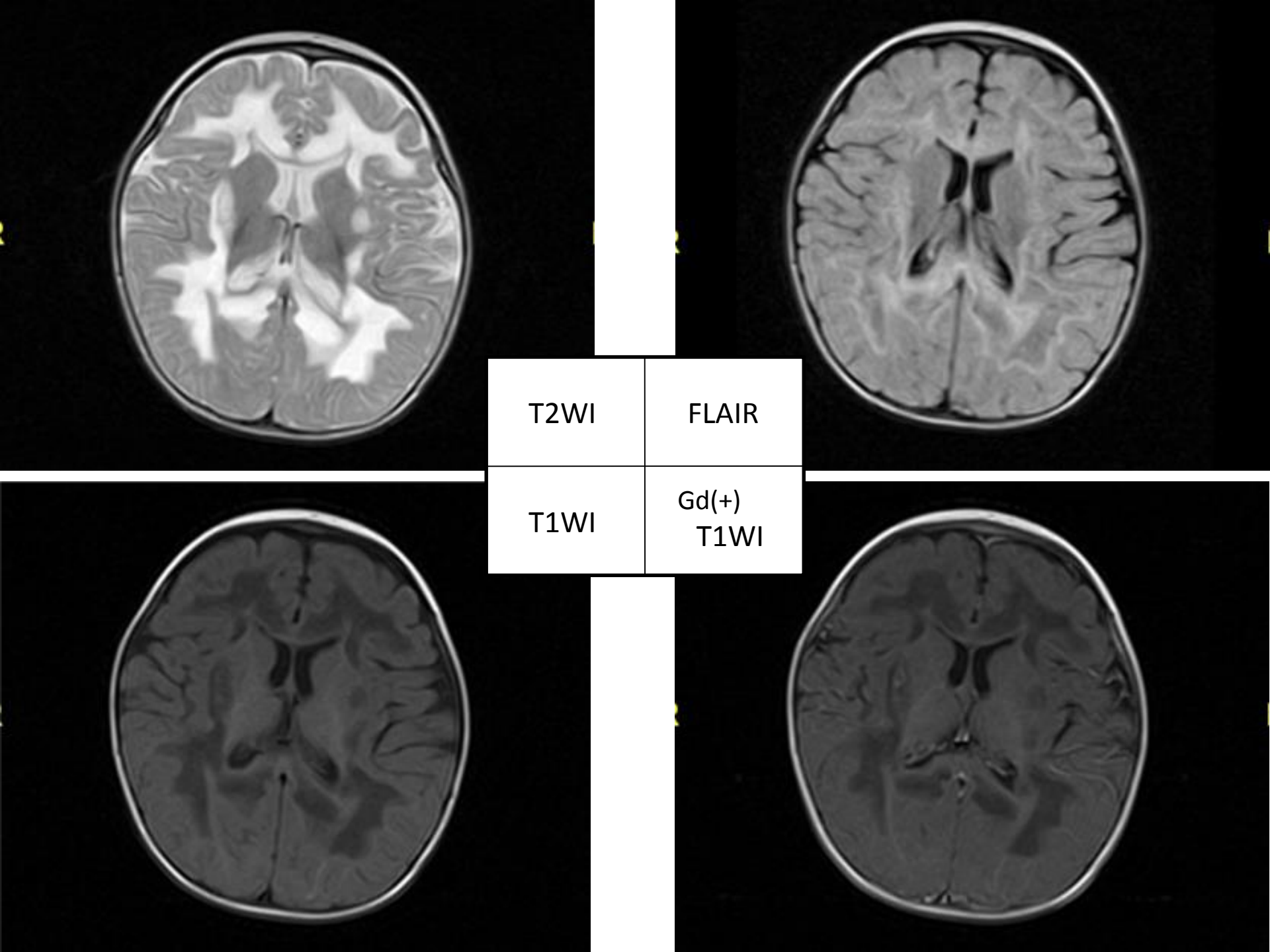
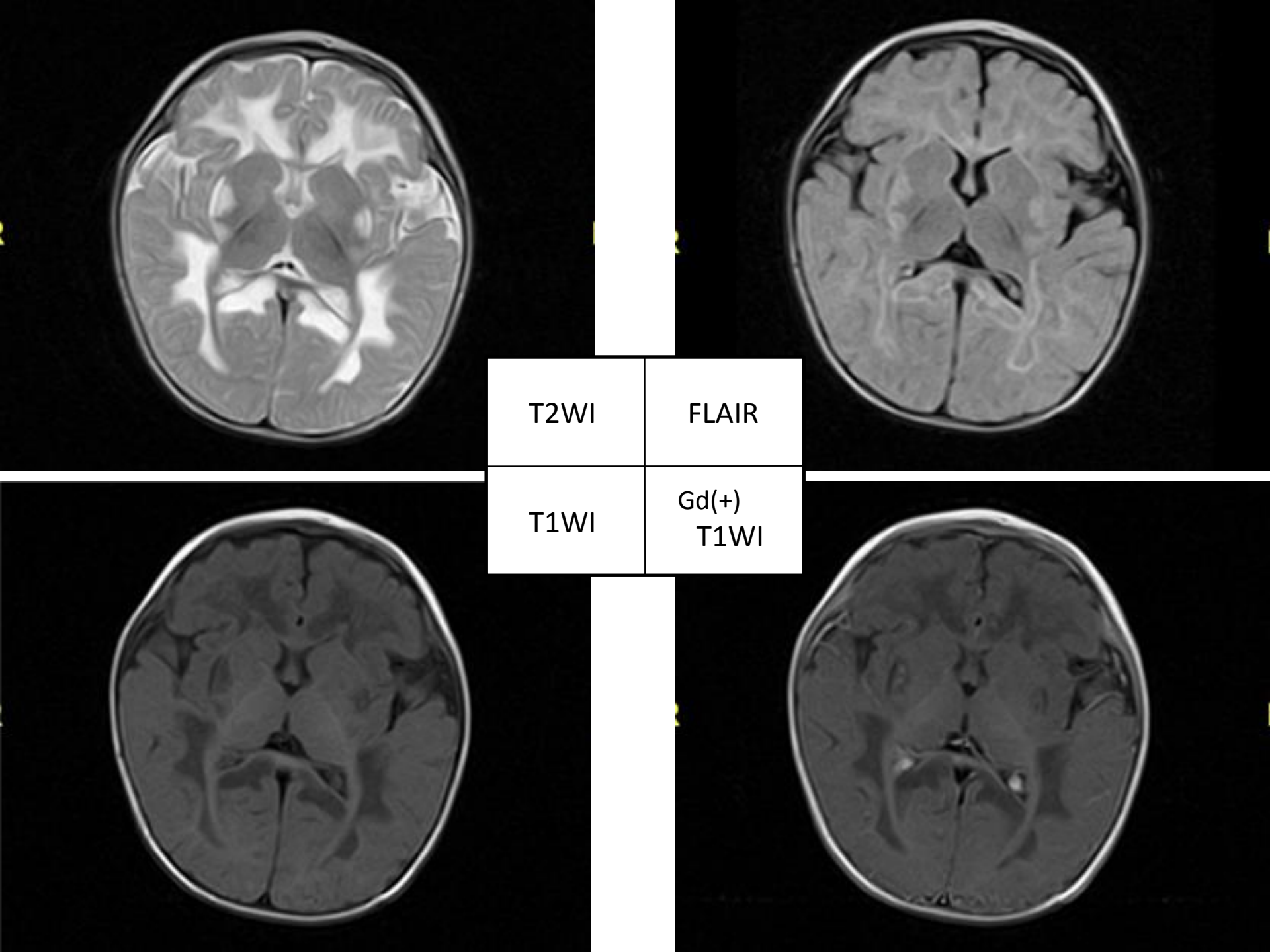
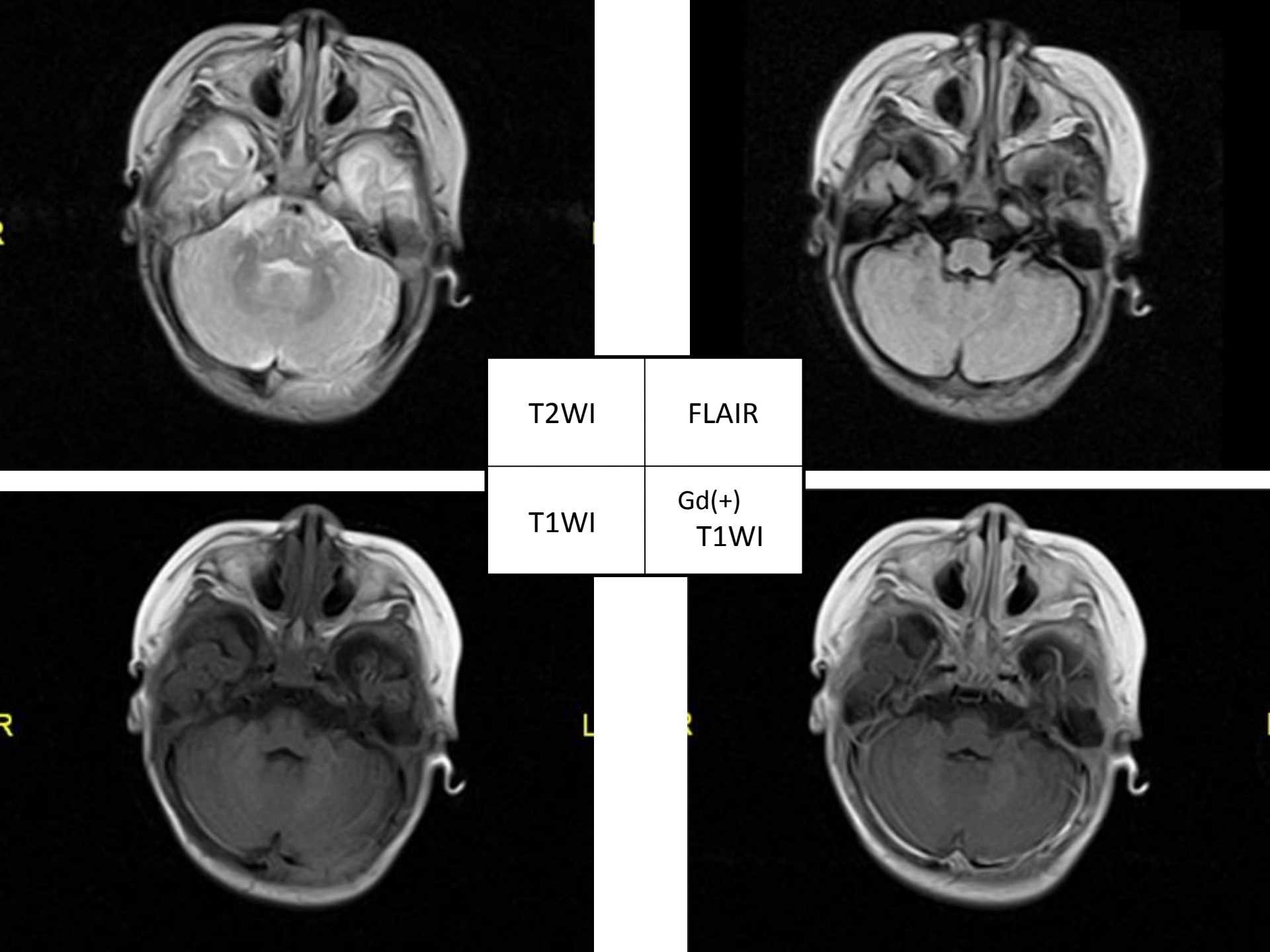
Clinical History

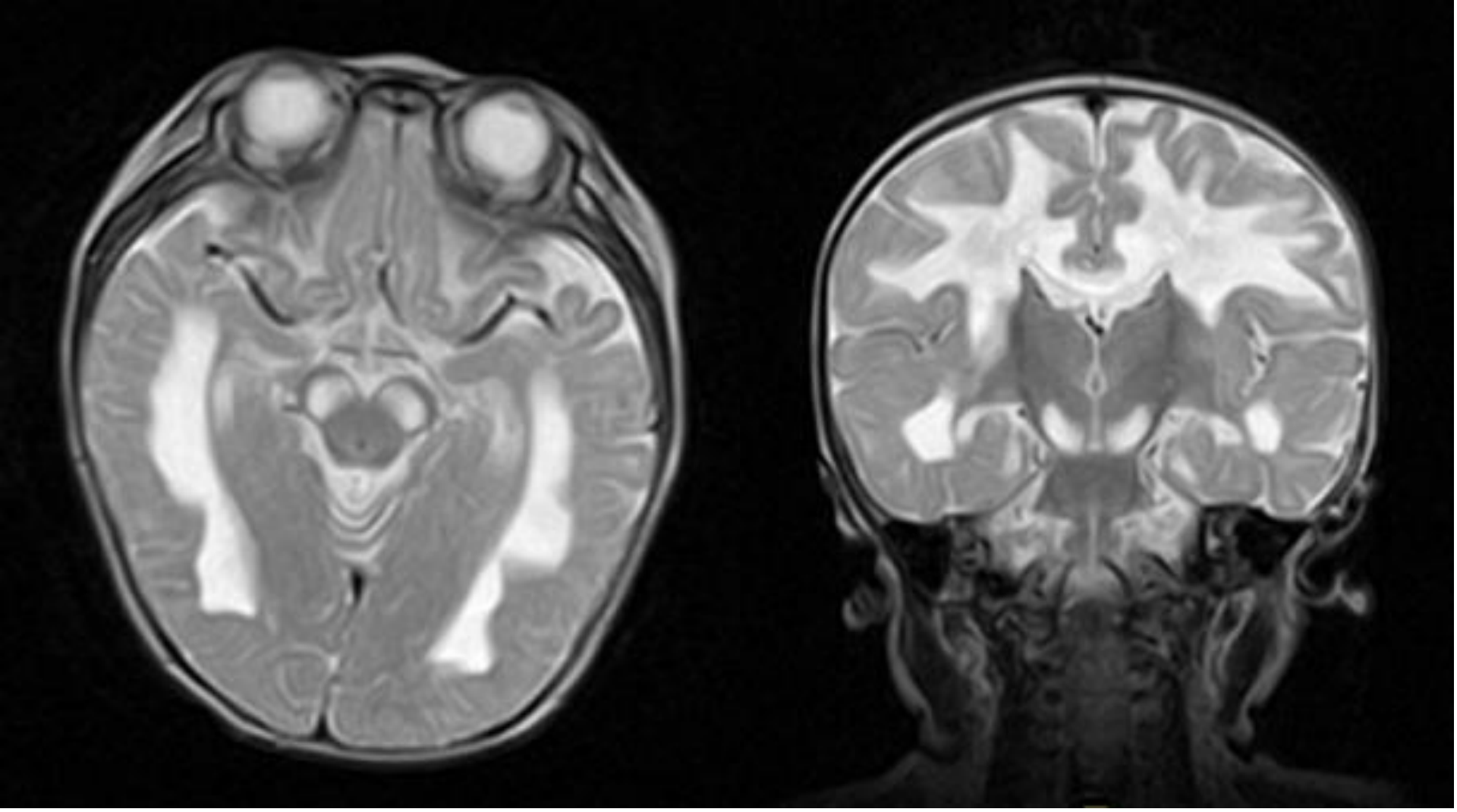
- Patient : 9 month-old girl
- Chief Complain : Regression and Myoclonus
- History of Present Illness :
  - No perinatal abnormality
  - Regression and myoclonus appeared after 7 months of age.
  - She was hospitalized and MRI test was performed at 9 months of age.
- Familial History: nothing in particular


T2WI
FLAIR
T1WI
Gd(+)
T1WI

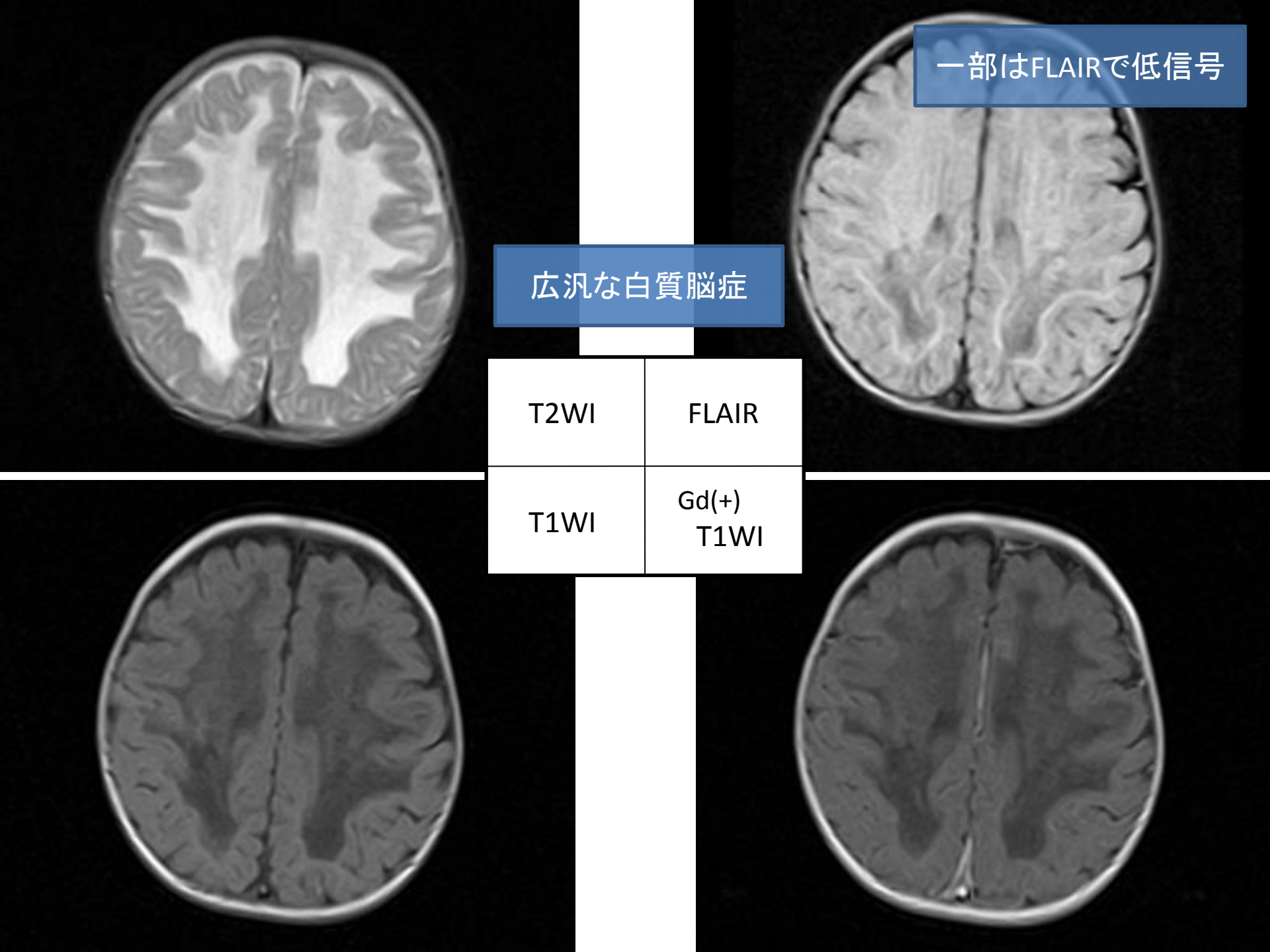

T2WI
FLAIR
T1WI
Gd(+)
T1WI

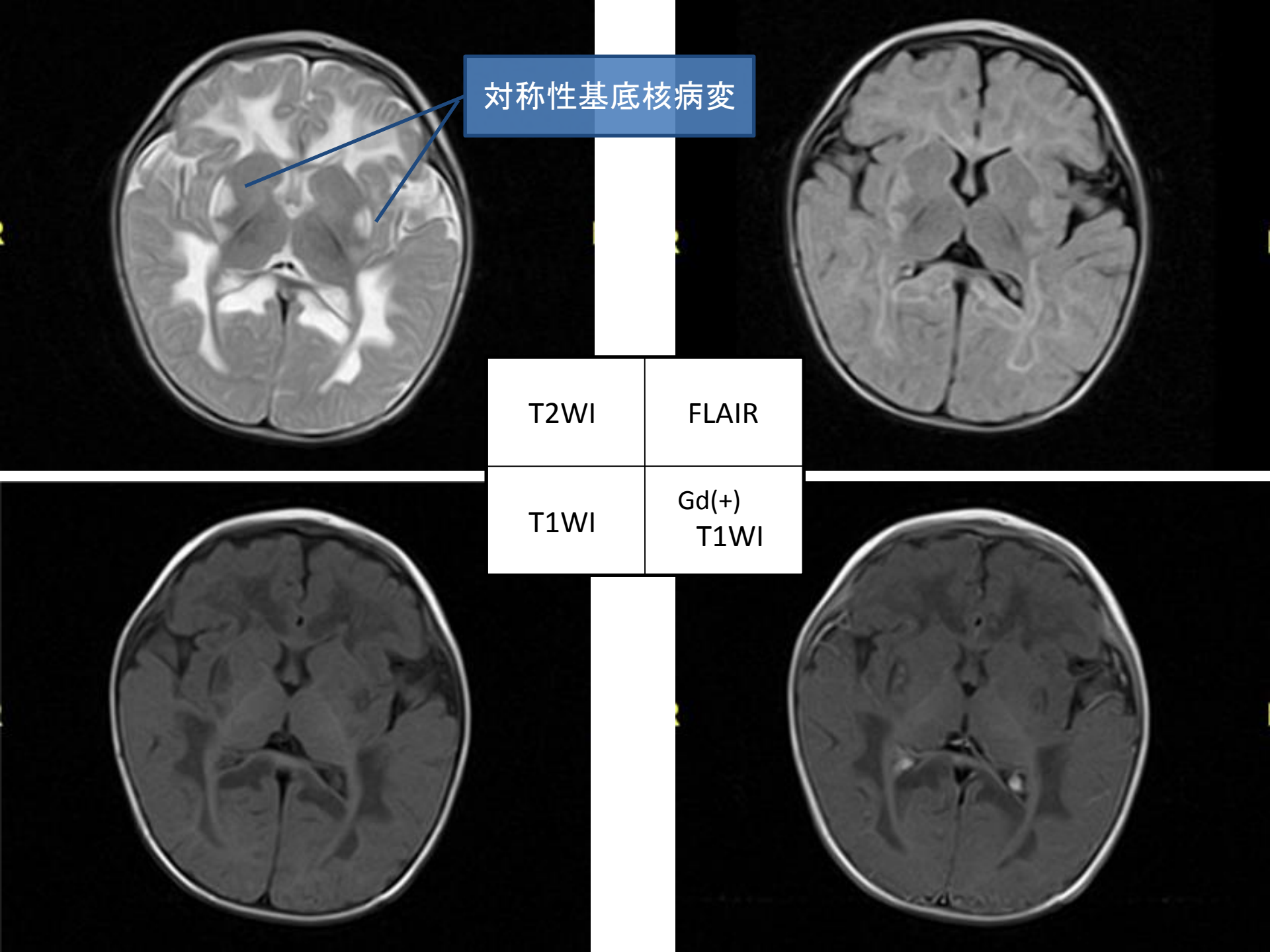

T2WI
FLAIR
T1WI
Gd(+)
T1WI

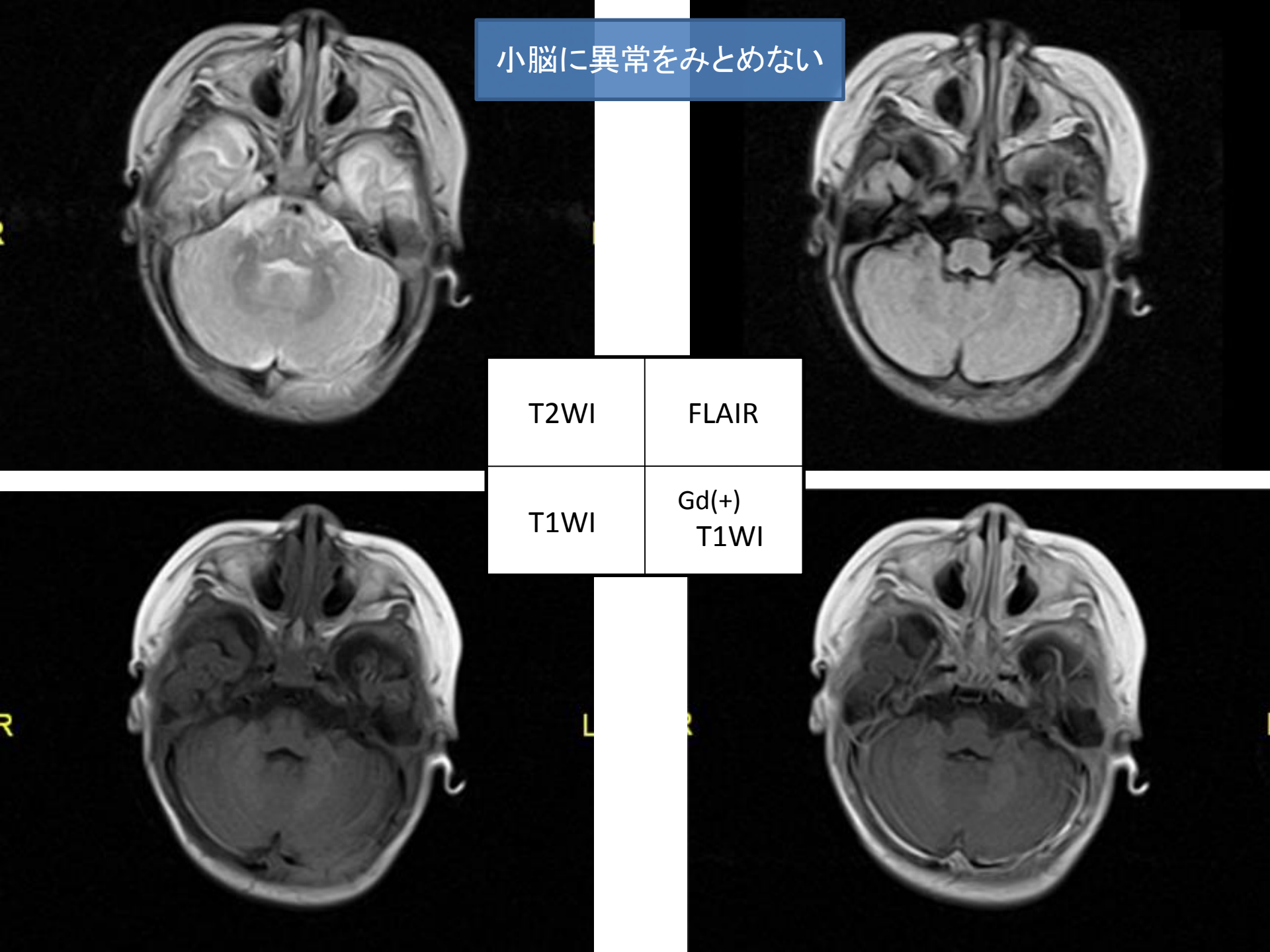

T2WI
FLAIR
T1WI
Gd(+)
T1WI

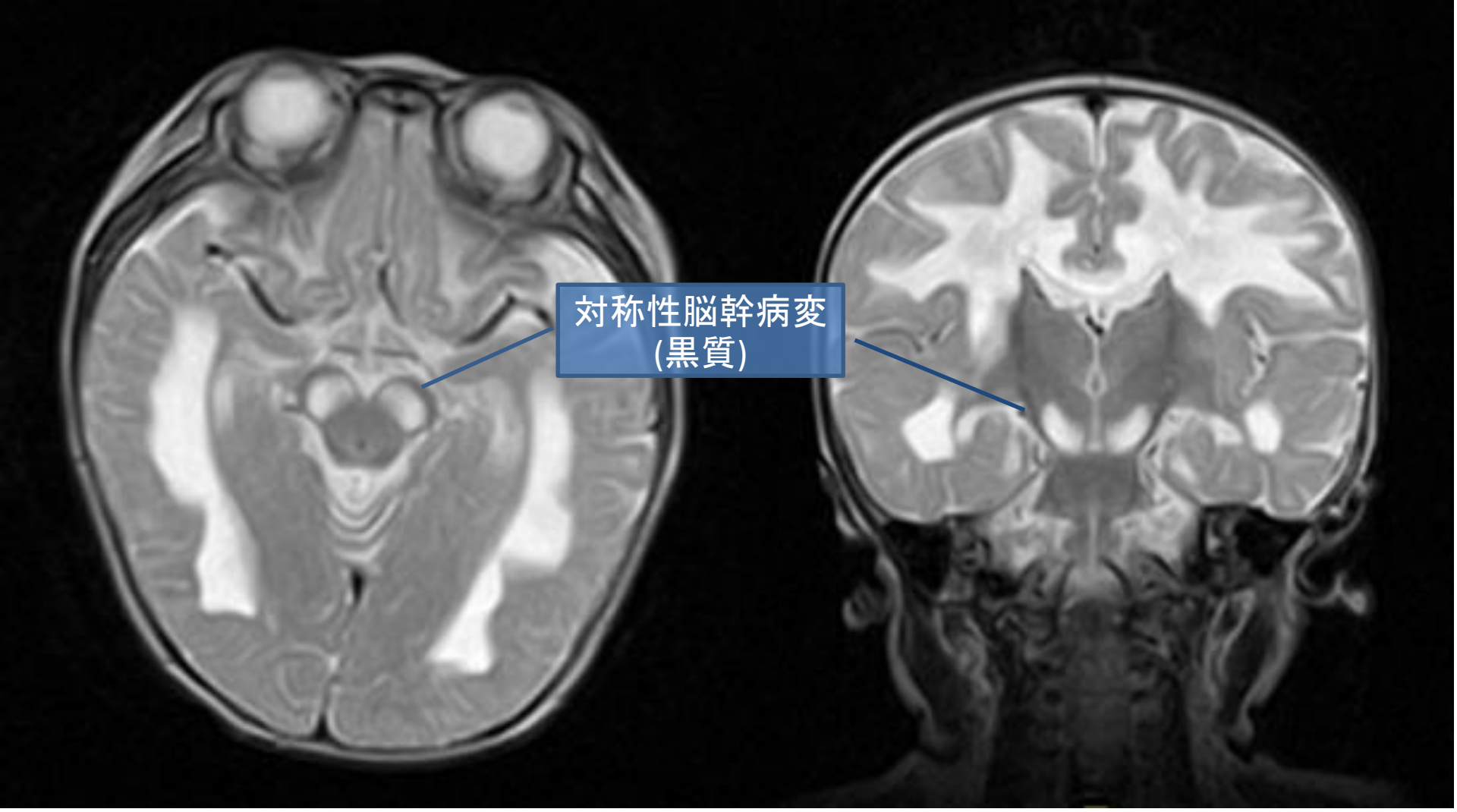


T2WI


一部はFLAIRで低信号
広汎な白質脳症
T2WI
FLAIR
T1WI
Gd(+)
T1WI


对称性基底核病变
T2WI
FLAIR
T1WI
Gd(+)
T1WI


小脳に異常をみとめない
T2WI
FLAIR
T1WI
Gd(+)
T1WI



T2WI

診断
Mitochondrial Disorder (complex Ⅰ deficiency)

培養皮膚線維芽細胞におけるComplexⅠの活性低下から診断された

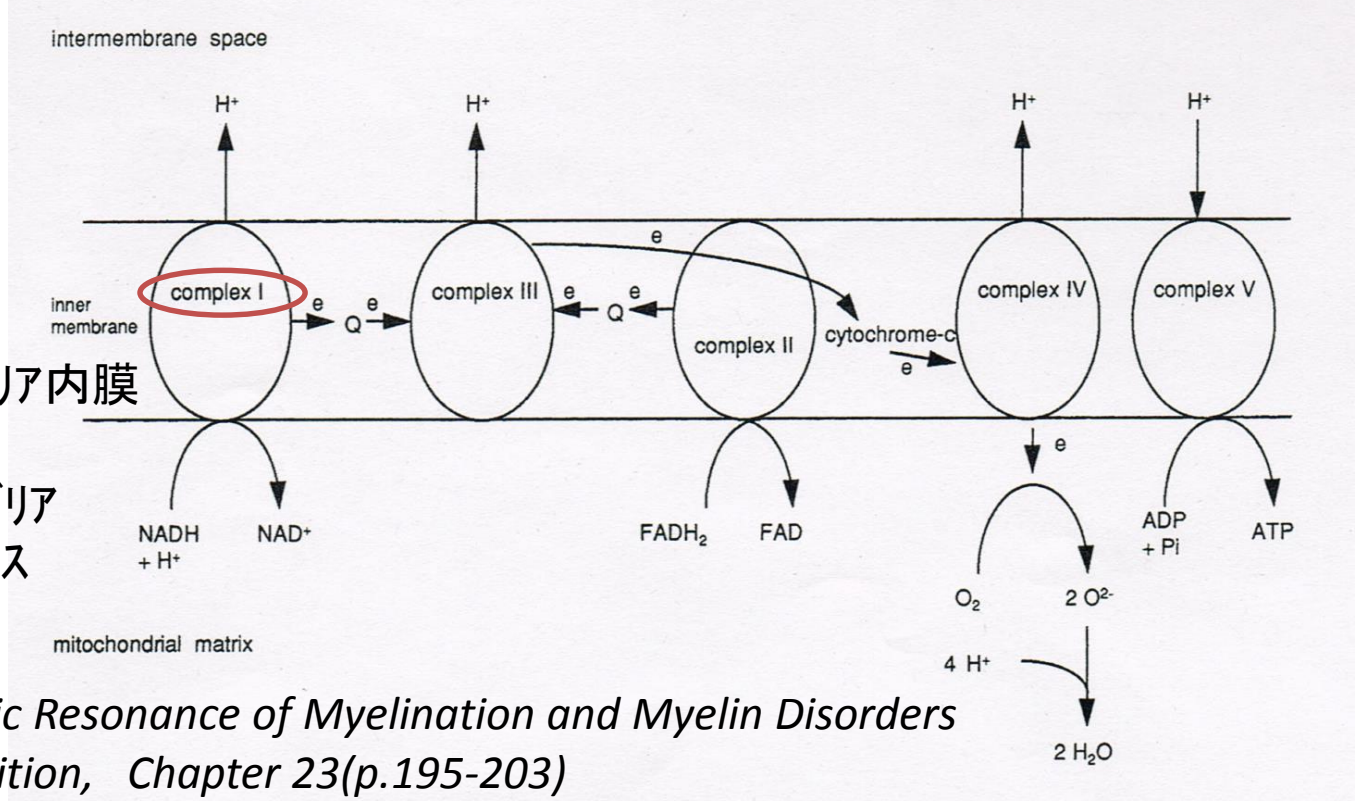
% of normal 28.3

CS ratio(%) 15.6

CoⅡ ratio(%) 21.5

complex I

- 酸化的リン酸化に関与する酵素複合体の一つ
- mtDNA やnDNAにコードされるcore subunitと複数の集合因子よりなる
- isolated complex Ⅰ deficiencyは呼吸鎖異常の最大の原因



Magnetic Resonance of Myelination and Myelin Disorders Third Edition, Chapter 23(p.195-203)

脳MRI

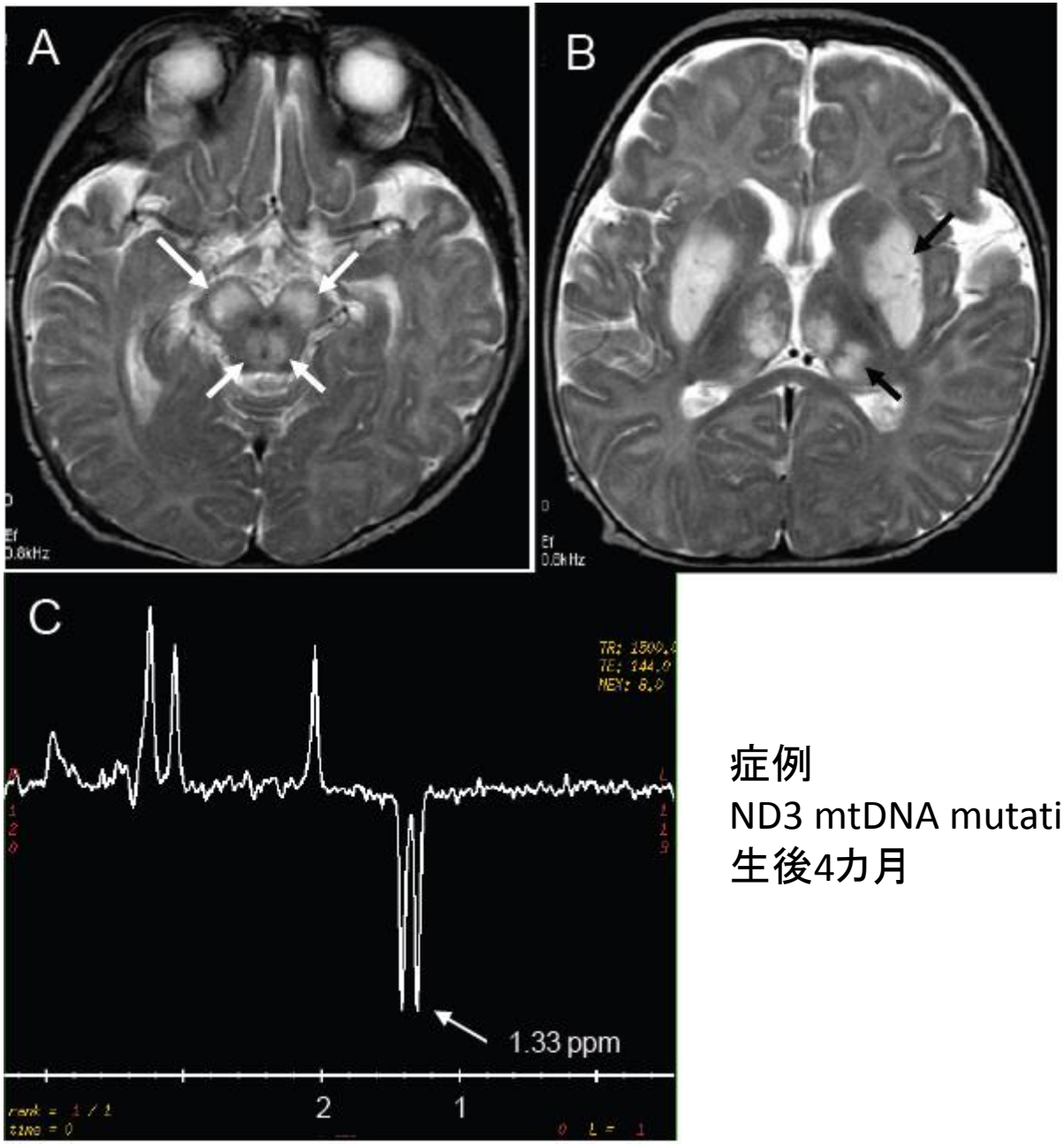
-30例のcomplex Ⅰ deficiency での検討-

A common pattern of brain MRI imaging in mitochondrial diseases with complex Ⅰ "Journal of Medical Genetics 48, 1 (2010) 16“ As Lebre, PhD et al.

- 脳幹病変(100%):左右対称
- 基底核:被殻(77%)、淡蒼球(53%)、尾状核(37%)
- 黒質、中脳水道周囲灰白質、乳頭視床路、脊髄視床路、内側毛帯、内側縦束に病変が多い
- 小脳病変(45%):5歳以下では見られない

※下線は本例で見られたもの

- MRS:乳酸のピークが全例(10/10)で見られた。
- CT:基底核に石灰化は見られない(0/3)



症例
ND3 mtDNA mutation
生後4カ月

A common pattern of brain MRI imaging in mitochondrial diseases with complex Ⅰ
"Journal of Medical Genetics 48, 1 (2010) 16" As Lebre, PhD et al.

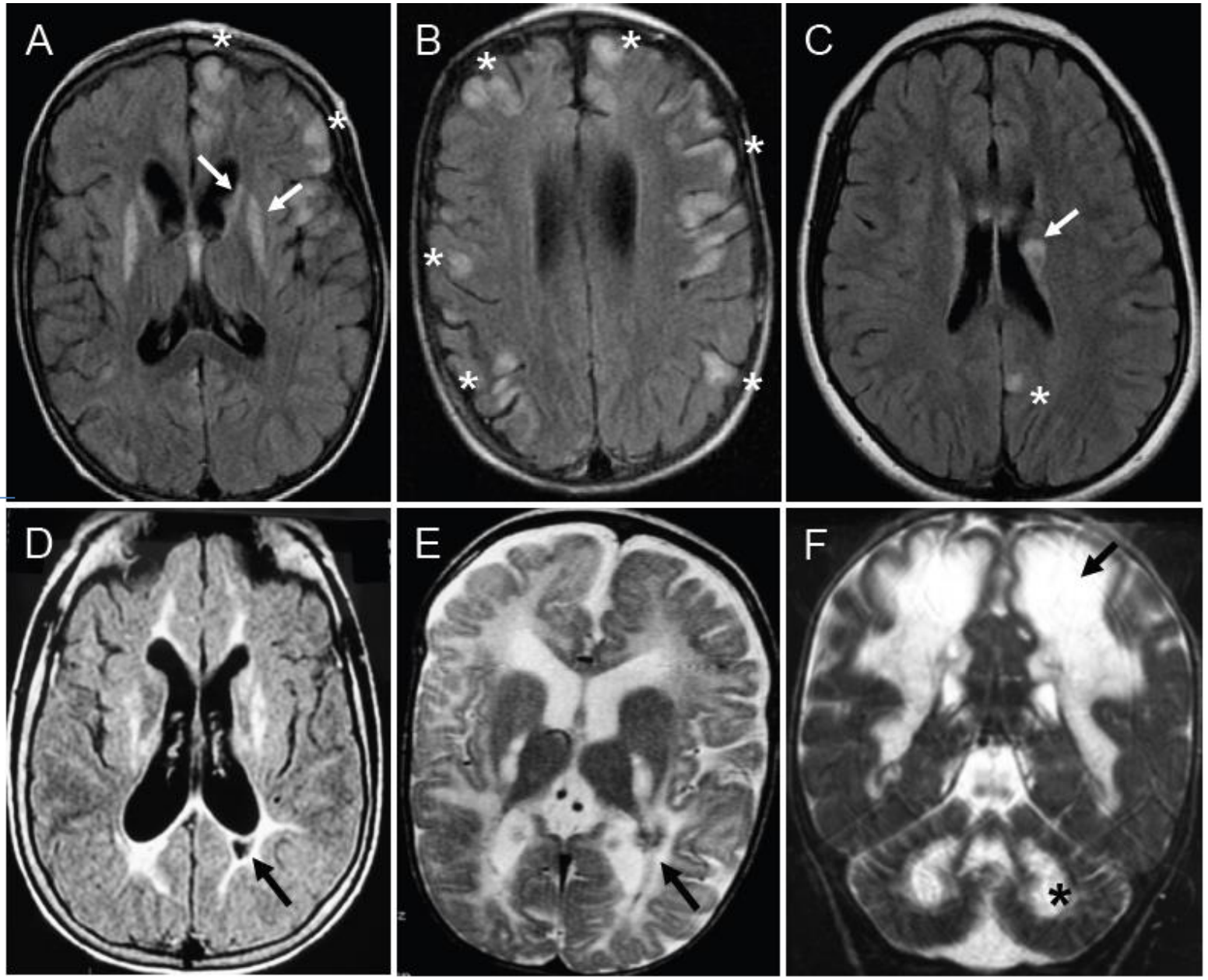
mtDNA mutation vs nDNA mutation

	mtDNA mutation(20例)	nDNA mutation(10例)
平均発症年齢	8.9歳	2.8歳
画像所見	•脳幹病変(100%) •テント上梗塞様病変(40%) •視床下核 •中脳水道周囲灰白質 •上丘 •尾状核病変 •小脳萎縮(5歳以上で)	•脳幹病変(100%) •深部白質を含む広範な白質脳症(50%) •特にNDUFS1 mutationでは壊死性病変が多い

A common pattern of brain MRI imaging in mitochondrial diseases with complex Ⅰ "Journal of Medical Genetics 48, 1 (2010) 16" As Lebre, PhD et al.

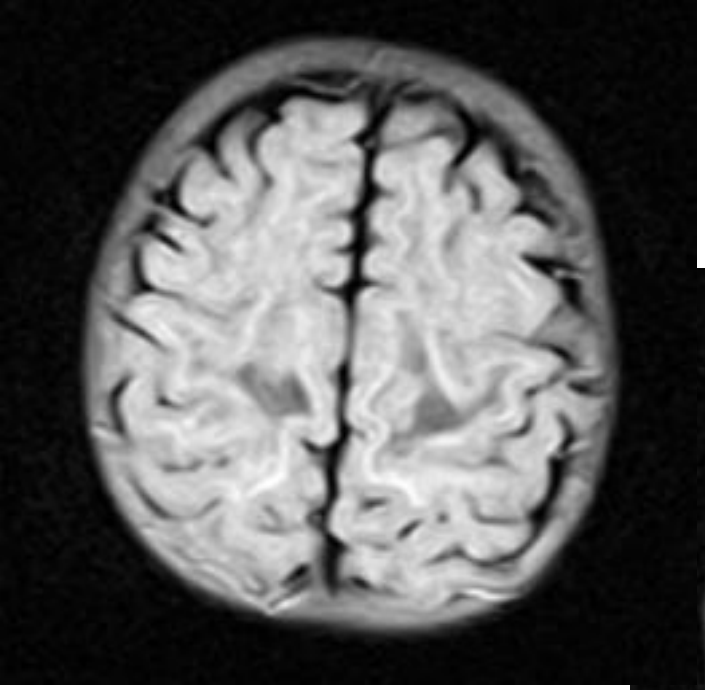
上段： mtDNA の変異

下段： nDNA の変異

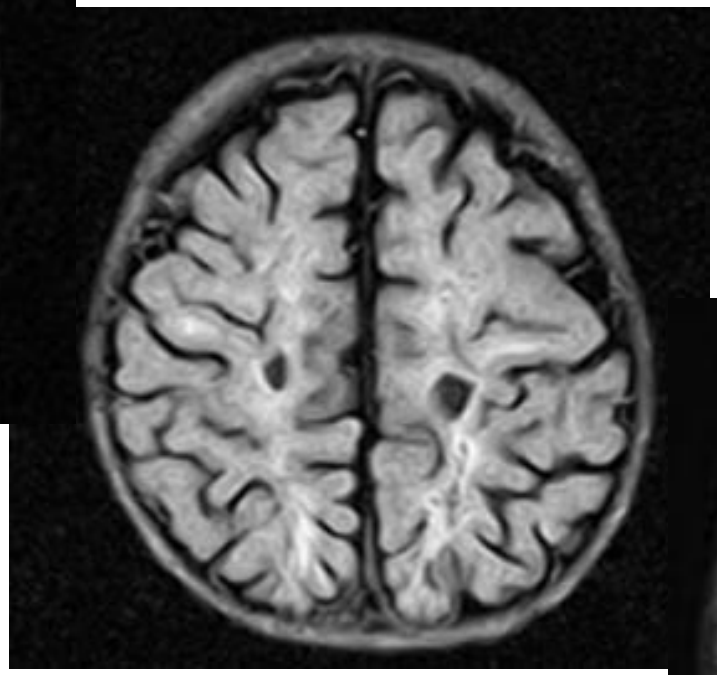


A common pattern of brain MRI imaging in mitochondrial diseases with complex Ⅰ
"Journal of Medical Genetics 48, 1 (2010) 16" As Lebre, PhD et al.

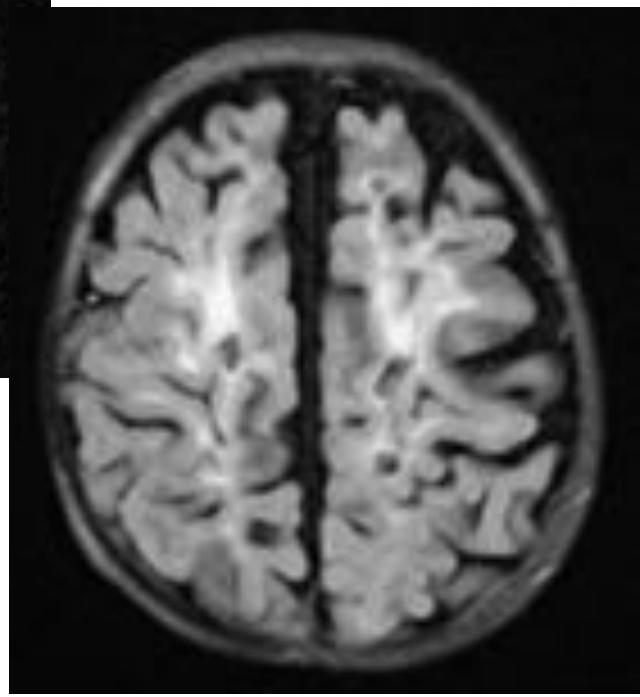
半卵円中心レベル
（FLAIR像)



2000/10/12
（生後9ヶ月)



2002/1/22
（生後24ヶ月)



2012/12/11
(12歳11ヶ月)

本例のその後の経過

Complex Ⅰ deficiencyの特徴

–年少で発症する
–脳幹病変の存在
–1つ以上の基底核病変を合併
–白質脳症or梗塞様病変を合併

A common pattern of brain MRI imaging in mitochondrial diseases with complex Ⅰ "Journal of Medical Genetics 48, 1 (2010) 16“ As Lebre, PhD et al.