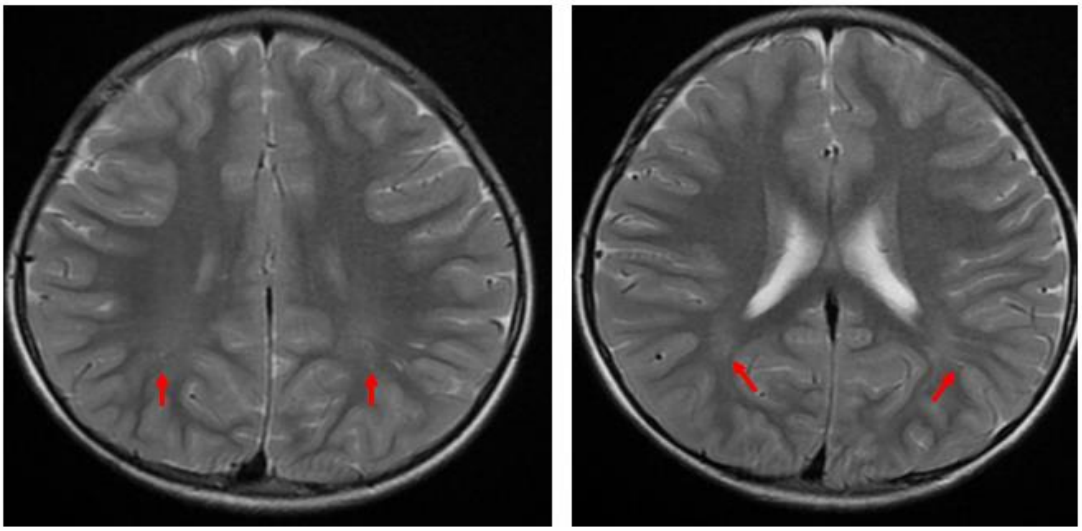
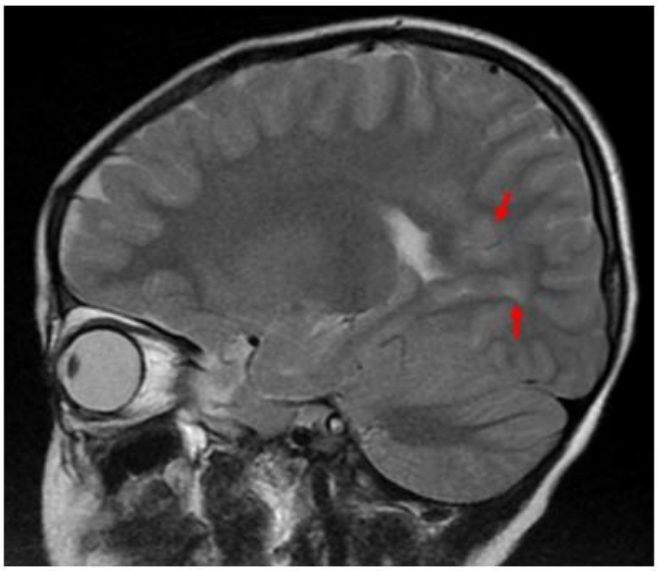
Case 418

白質形成不全



限局性の高信号域が脳室背側の連合野白質に認められる。
白質内に低吸収束を含んでいる。



T2WIの矢状断で斑状の高信号が頭頂葉と後頭葉の連合野に認められる。
内部に低吸収束を含んでいる。

Leukodystrophy：白質形成不全

白質＝軸索成分が2億から3億

二つに分類

・低形成 hypomyelinating
・脱髄性変化 demyelinating

神経細胞
遺伝子の欠損或いは異常配列でで本来利用されるべき或いは利用されない
リン酸化合物や長鎖脂肪酸が沈着
白質の電気信号の伝達の不具合が生じ、神経機能の低下を招来

Leukodystrophy：白質形成不全 Hypomyelinating

- 遺伝子異常により

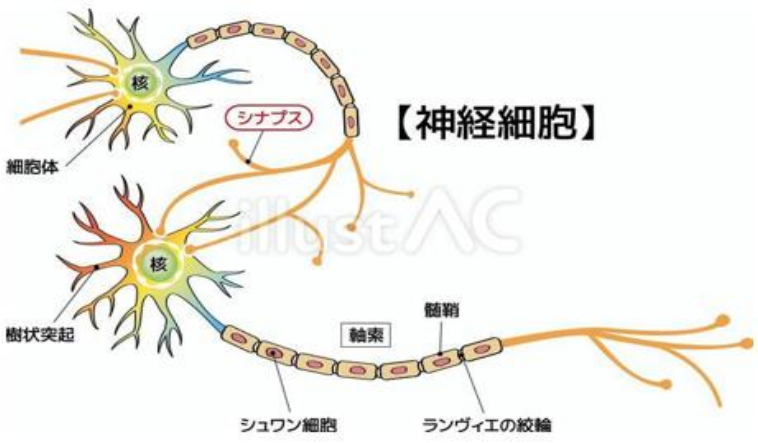
- structural myelin proteins, ミエリン構造蛋白の形成不良
- oligodendrocyte transcription factors,
- RNA translation
- lysosomal proteins

神経細胞：

電気信号受信
：樹状突起

電気信号送信
：軸索突起

軸索には髄鞘が付随
電気信号の高速に関与



髄鞘：oligodendrocyte から栄養補給

Leukodystrophy：白質形成不全

- X-linked adrenoleukodystrophy,
- Metachromatic leukodystrophy,
- Krabbe's disease,
- Pelizaeus–Merzbacher disease,
- Alexander's disease,
- Canavan disease,
- Aicardi–Goutières Syndrome

- これらは、特有の遺伝子異常の検索が進められている

Types of Leukodystrophies

	Genetic Inheritance	MRI Findings	CSF/Bloodwork Markers	Diagnostic Modalities	Other Key Features
1 X-linked Adrenoleukodystrophy	X linked	White matter changes Parieto-occipital involvement	Elevated VLCFA levels in blood	MRI VLCFA blood test	Adrenal gland involvement, adrenal insufficiency, neurodegeneration
2 Metachromatic Leukodystrophy	Autosomal Recessive	Periventricular white matter changes Corpus callosum involvement	Reduced arylsulfatase A levels in blood or urine	MRI Arylsulfatase A assay	Motor and cognitive deterioration
3 Krabbe's Disease	Autosomal Recessive	Periventricular and subcortical changes Involvement of the thalamus and globus pallidus	Elevated CSF protein levels	MRI Nerve biopsy	Peripheral neuropathy, optic atrophy
4 Pelizaeus-Merzbacher Disease	X linked	Diffuse white matter abnormalities Involvement of the corpus callosum	Low levels N-acetyl Aspartate	MRI Genetic testing	Nystagmus, tremor, ataxia
5 Alexander's Disease	Sporadic or Autosomal Dominant	White matter changes Frontal predominance Basal ganglia involvement	Elevated GFAP levels in CSF	MRI Genetic testing	Macrocephaly, seizures
6 Canavan Disease	Autosomal Recessive	Diffuse white matter abnormalities Subcortical U-fiber involvement	Elevated NAA levels in urine	MRI Genetic testing	Macrocephaly, severe motor impairment
7 Aicardi-Goutières Syndrome	Autosomal Recessive	Periventricular and deep white matter abnormalities Brain calcifications*	Elevated levels of interferon-alpha and other cytokines	MRI Genetic testing	Microcephaly, spasticity, seizures

* Brain calcification are better visualized by cranial CT studies

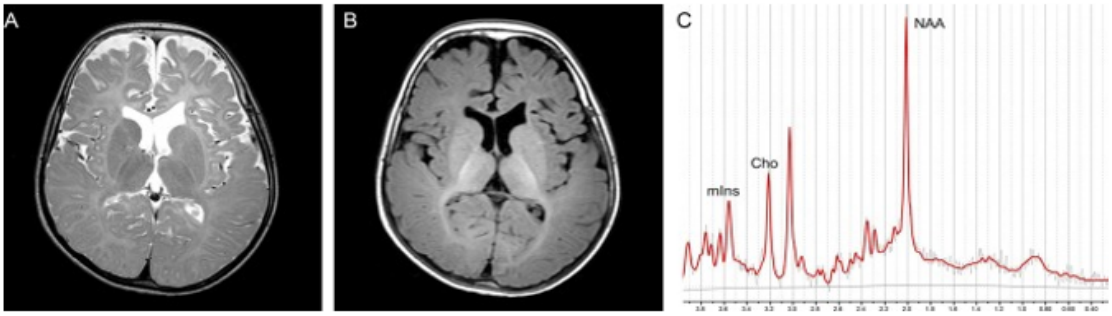
白質不全症の診断にはまず、MRIとそれに引き続く検査が示されているが
MRI　spectroscopyが有用

Leukodystrophy：白質形成不全
MRI：画像とMRI spectroscopy

- **画像：T2WIで高信号**

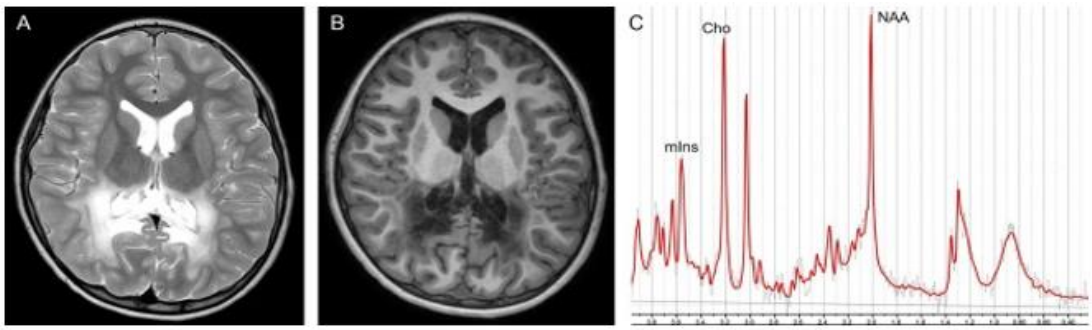
- **MRI　spectroscopy：oligodendrocyteやglia cell の代謝成分を評価**

- **遺伝子異常**



低形成型

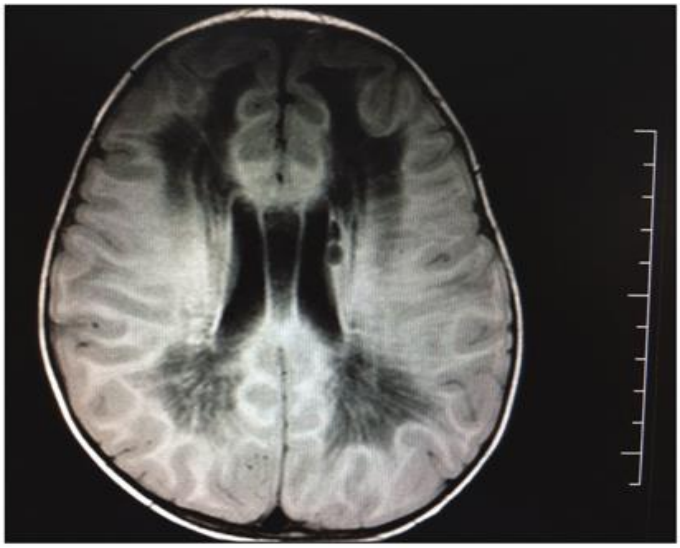
Elementary school boy with Pelizaeus–Merzbacher disease with *PLP1* duplication, a representative hypomyelinating leukodystrophy.
T2-weighted image (A) showing mild diffuse hyperintensity in the white matter, with hypointensity in the posterior limb of the internal capsule indicating myelination. T1-weighted image (B) showing the white matter having nearly equal signal to the cortex, with hyperintensity in the posterior limb of the internal capsule and optic radiation reflecting myelination. MRS of the left white matter (C) appears normal, with quantitative analysis showing normal NAA, decreased Cho, and increased mIns.
NAA, *N*-acetyl aspartate; Cho, choline; mIns, myo-inositol.

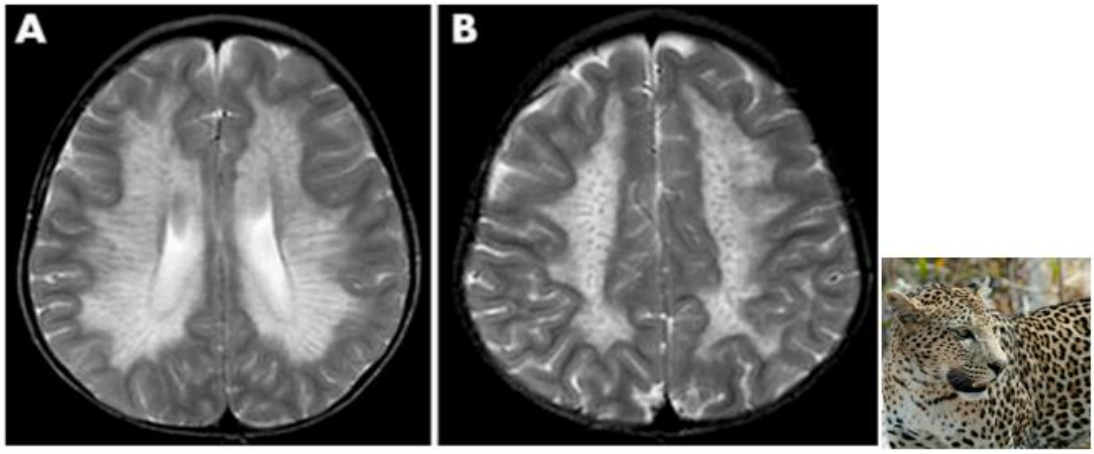


脱髄型

Elementary school boy with adrenoleukodystrophy, a representative demyelinating leukodystrophy.
T2-weighted (A) and T1-weighted (B) images showing prominent hyperintensity and hypointensity in bilateral occipital periventricular white matter. MRS (C) showing markedly reduced NAA.
NAA, *N*-acetyl aspartate.

低形成型

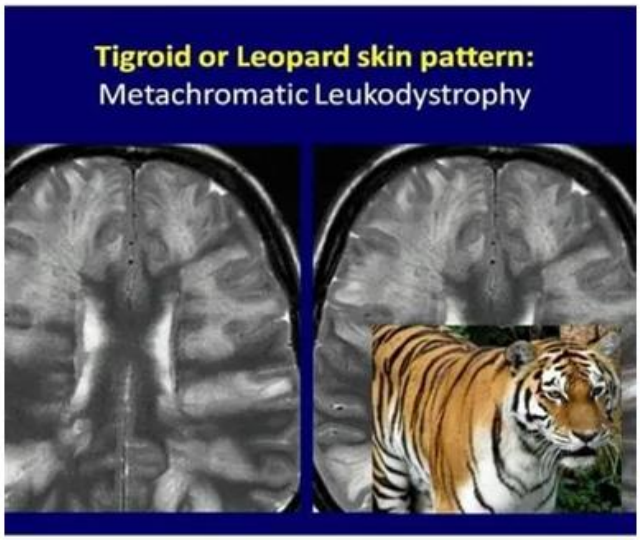


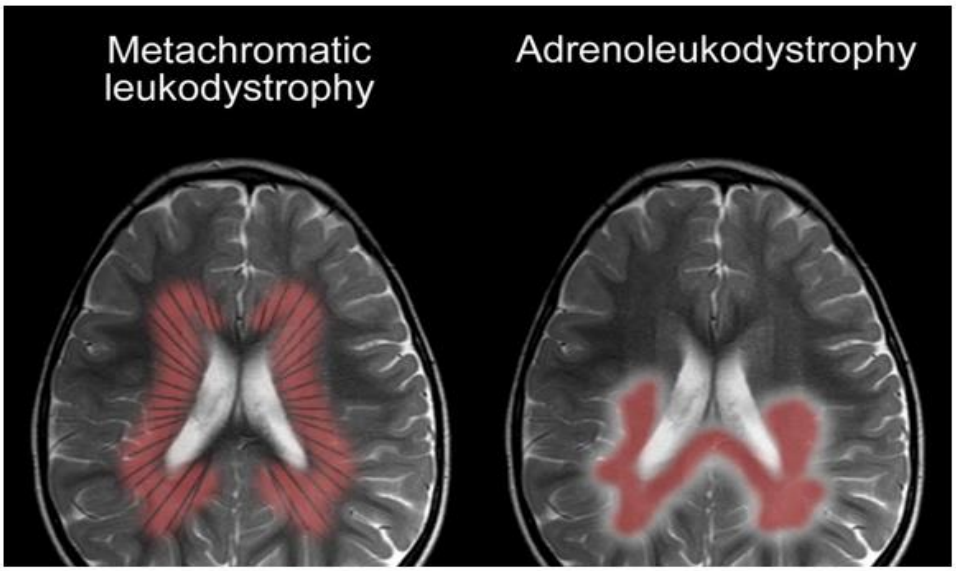


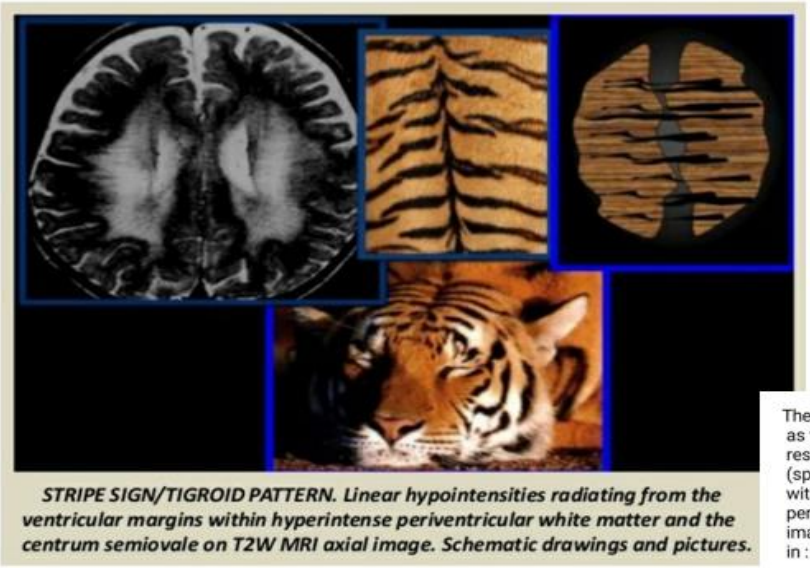
低形成型

metachromatic leukodystrophy：白質にSulphatides リン酸代謝物の沈着。

低形成型







STRIPE SIGN/TIGROID PATTERN. Linear hypointensities radiating from the ventricular margins within hyperintense periventricular white matter and the centrum semiovale on T2W MRI axial image. Schematic drawings and pictures.

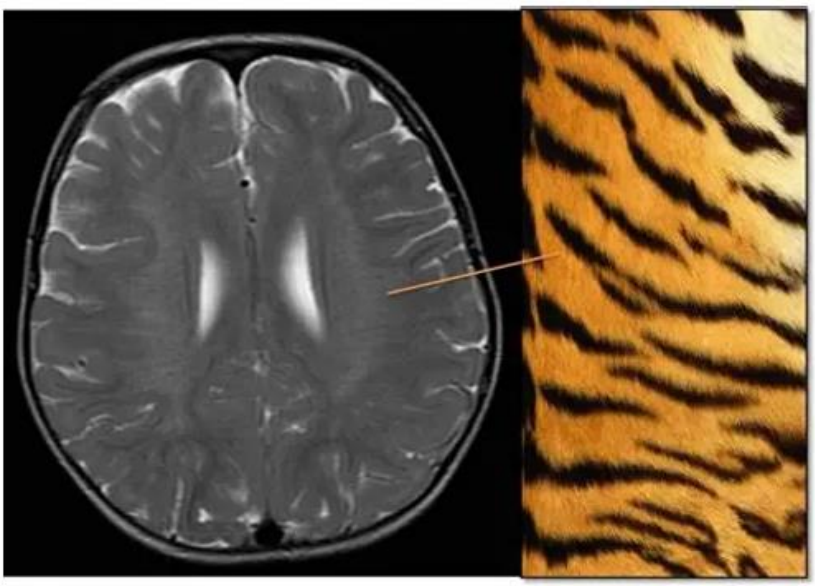
The Leopard skin sign (also known as tigroid pattern or stripe sign) results from dark-spots or stripes (spared perivascular white matter) within bright demyelinated periventricular white matter on T2W images. It is characteristically seen in :

a) Metachromatic leukodystrophy

b) Pelizaeus-Merzbacher disease

c) Autosomal recessive spastic ataxia of Charlevoix

低形成型



metachromatic leukodystrophy

Leukodystrophy：白質形成不全

- MRIが有用：T2WIで高信号
- 白質のT2WIで高信号：子供や若年者発症

- 低形成型：T2WIで比較的淡い高信号、内部に低信号のライン(軸索保持)
  （Tigroid, Leopard line ）
- 髄鞘型：T2WIで高信号、皮質下白質にまで及ぶ

- 問題点
  高齢者：加齢性のT2WI,FLAIRの高信号と類似
  Glymphatic systemの異常か脱髄の変化か不詳
  ：電気信号の伝達機能低下を示唆しているのかもしれない

English page

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