

CASE 2

15 Y/O BOY

Brief history

- **Past history**

Denied

- **Chief complain**

Recurrent right facial palsy and right upper limbs weakness for one day

Imaging studies

Brain CT (no C) (2018.05.12)

Brain MRI (+ C) (2018.05.14)

Imaging findings

✓ CT :

Equivocal density of splenium (corpus callosum)

✓ MRI :

Symmetrical and bilateral hyperintensity changes in bilateral motor tracts, splenium of the corpus callosum and parietal white matter

Imaging findings



Differential Diagnoses

- Hypoglycemia
- Acute disseminated encephalomyelitis (ADEM)
- HSV encephalitis

Other findings

- ✓ Lumbar puncture: normal
- ✓ Plain film: **Pec cavus**
- ✓ Neurological conductive study :
 - **diffusely demyelinating sensorimotor polyneuropathy**
 - **sudomotor dysfunction** in lower limbs
 - **Peripheral neuropathy** in somatosensory evoke potential

Differential Diagnoses

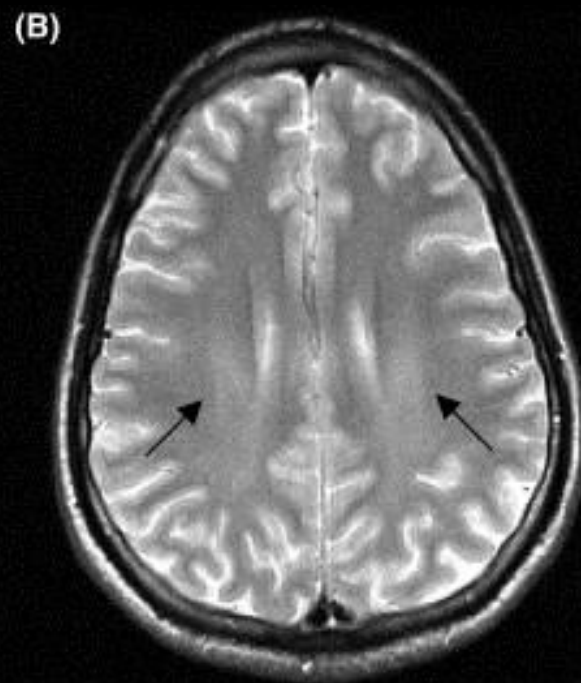
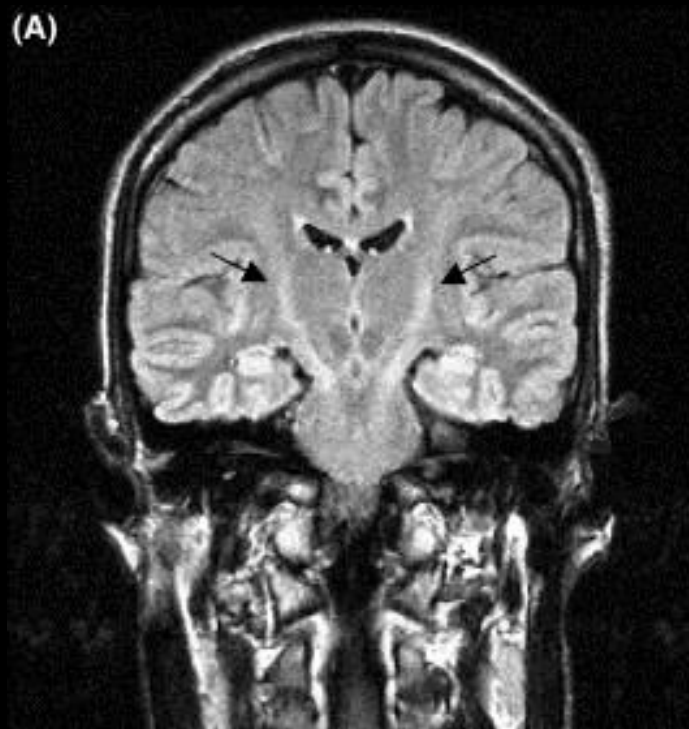
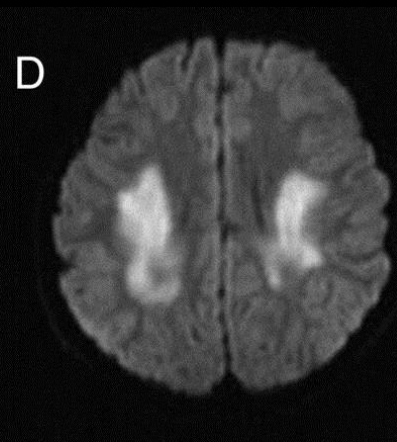
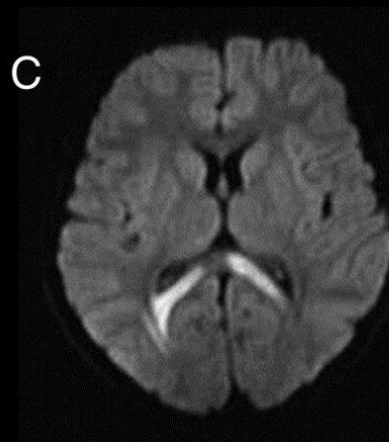
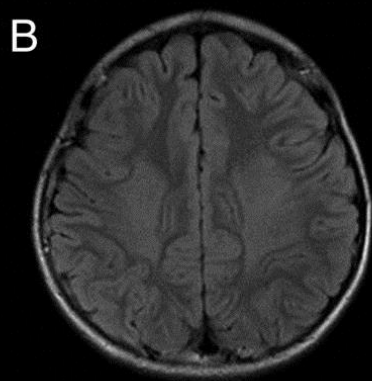
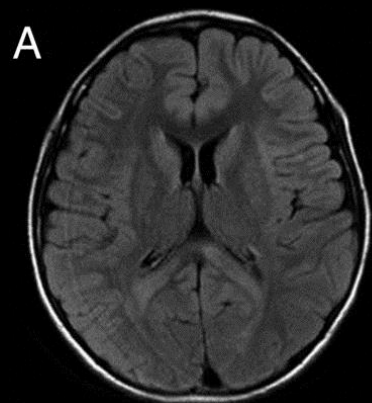
- Hypoglycemia
- Acute disseminated encephalomyelitis (ADEM)
- HSV encephalitis
- **X-linked Charcot-Marie-Tooth disease (CMTX1)**

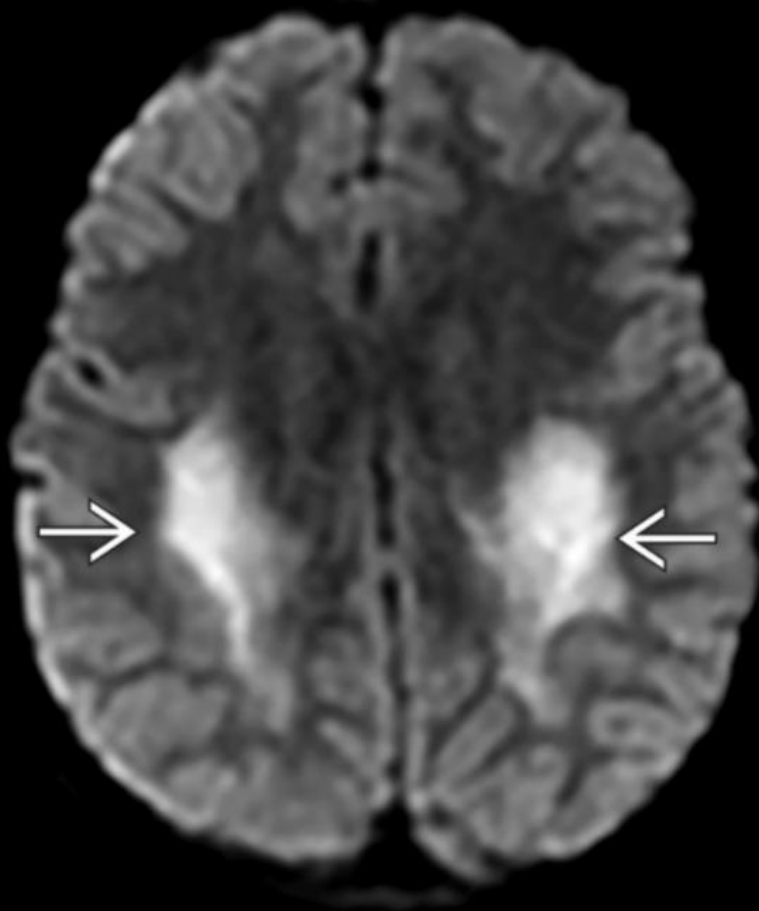
X-linked Charcot-Marie-Tooth disease

- X-linked CMT (CMTX), CMTX type 1 (CMTX1)
- **Heterogeneous hereditary peripheral sensory and motor neuropathy**; 2nd most common CMT variant after CMT1A
- Mutations in **gap junction beta 1 gene (*GJB1*)** on chromosome Xq13.1 coding for **connexin-32**
- **Acute transient encephalopathy**
- **Transient motor weakness**
- **Chronic distal leg muscle wasting $\pm$ weakness of ankle flexion**
- **Pes cavus foot deformity**

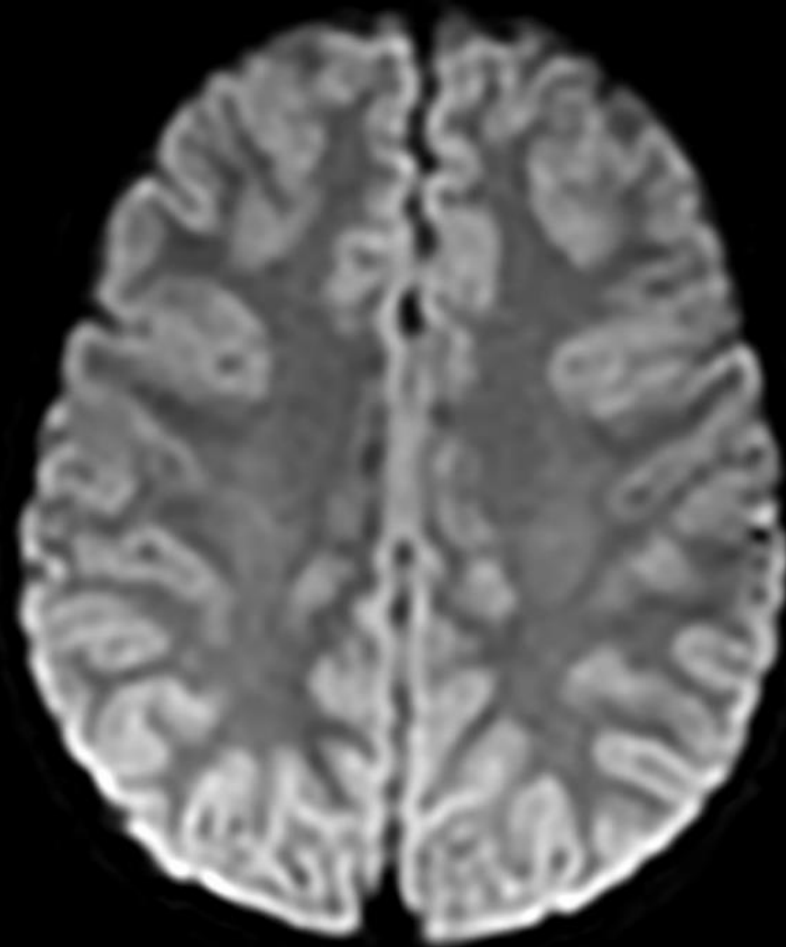
X-linked Charcot-Marie-Tooth disease

- **NECT :**
lesions normal (undetectable) to **slightly hypodense**
- **MRI :**
 - **confluent symmetric white matter T2-hyperintense lesions** in posterior centrum semiovale, splenium
 - acute lesions show **diffusion restriction**
 - spares subcortical U-fibers
 - **no lesion enhancement**
- White matter lesions transient; improvement or complete resolution common





Acute phase



1 month later