

34th Neuroradiology workshop 3-7

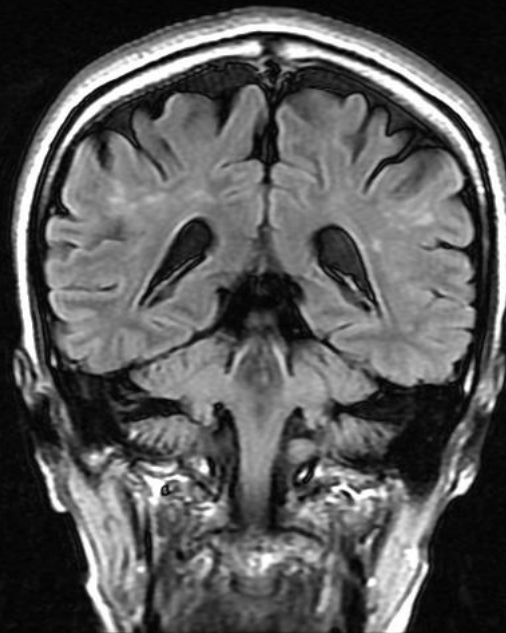
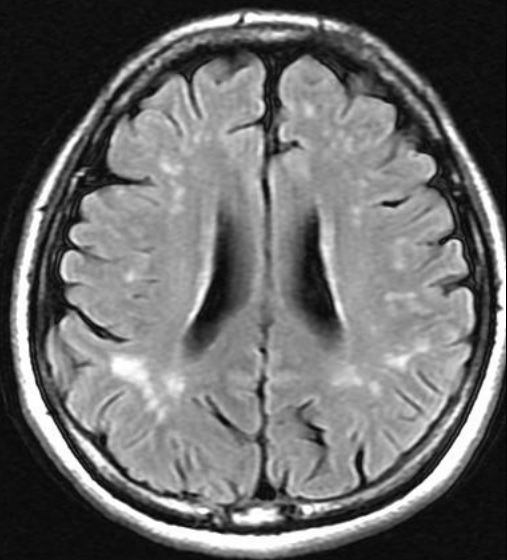
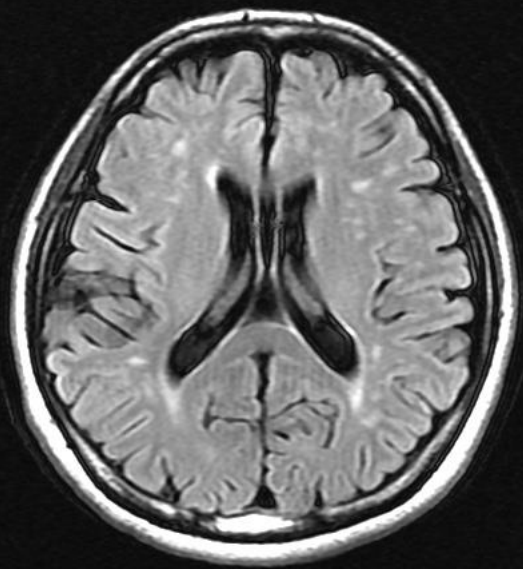
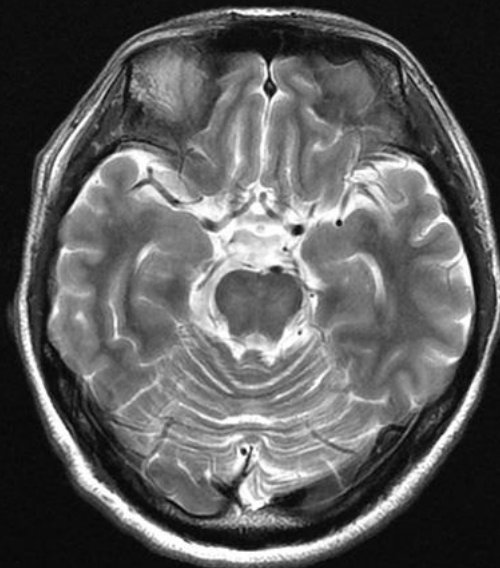
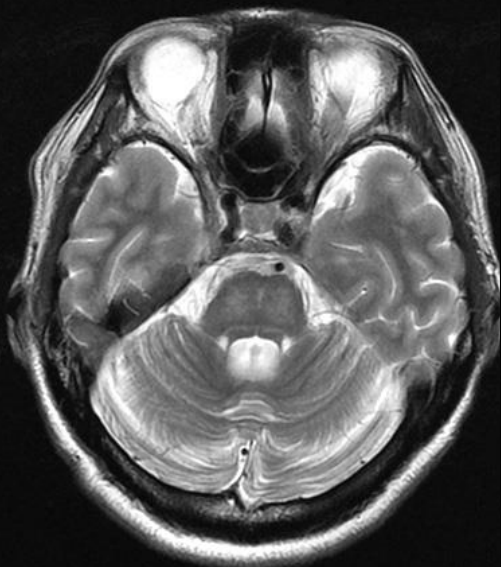
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Case

- A woman in her mid-40s
- Unsteadiness/difficulty in gait for 2 years
- Difficulties in speech and writing developed recently
- Her father and uncle also suffer from similar symptoms

Neurological exam

- Nose-finger-nose test: awkward
- Dysdiadochokinesia (+)
- No pathological reflexes



Diagnosis please

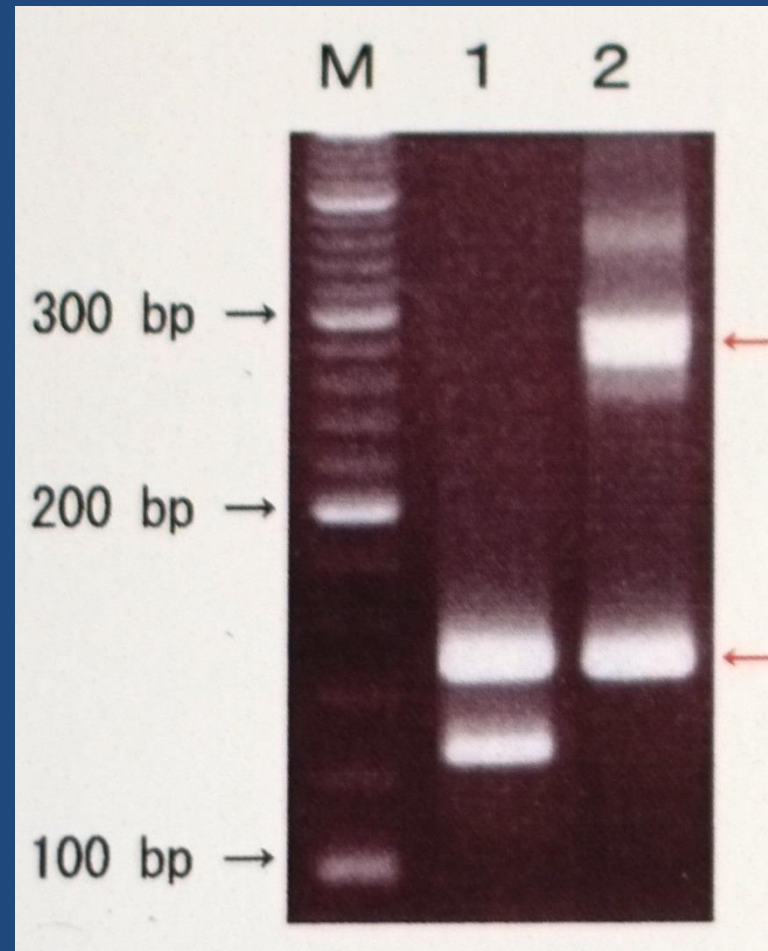
MR imaging findings

- Atrophy of the cerebellum, superior cerebellar peduncles, midbrain (81 mm²) and pons (416mm²)
- Patchy hyperintense lesions in the pontine base and deep cerebral white matter on FLAIR and T2-weighted images

- Positive family history and clinical and MR findings suggest DRPLA.

PCR analysis for DRPLA

- Number of expanded CAG repeats was 61 (normal range, 8 – 25 repeats)
 - M: size marker
 - 1: normal control
 - 2: sample of this case



Dentatorubral-Pallidoluysian Atrophy Naito-Oyanagi Disease

- Autosomal-dominant cerebellar ataxia characterized by ataxia, choroathetosis and dementia in adults, and ataxia, myoclonus, epilepsy and progressive intellectual deterioration in children. Clinical presentation varies depending on the age of onset.
- Histopathology: combined degeneration of the dentatorubral and pallidoluysian systems; diffusely decreased staining for myelin of the cerebral white matter
- CAG repeat expansion disease; abnormal expansion of unstable trinucleotide (CAG) repeats in atrophin-1 gene located on chromosome 12p13

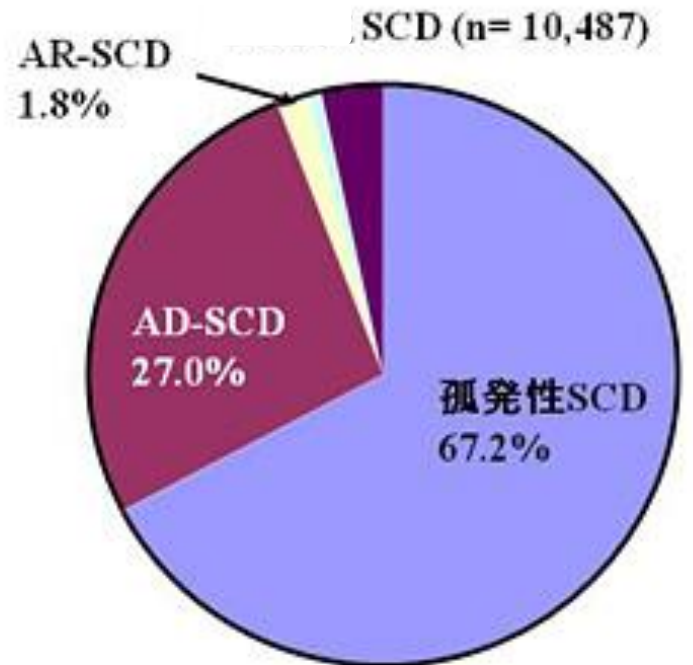
Clinical subtype of DRPLA

	Juvenile (≤ 20 years)	Early adult (20–40 years)	Late adult (≥ 40 years)
Epileptic seizure	+++	+	–
Myoclonus	+++	+	–
Dementia	+++	+++	+
Cerebellar ataxia	+	+++	+++

- Adult-onset (over age 20)
 - Progressive disorder of ataxia, choreoathetosis, and dementia
- Juvenile-onset (under age 20)
 - Ataxia, myoclonus, epilepsy and progressive intellectual deterioration

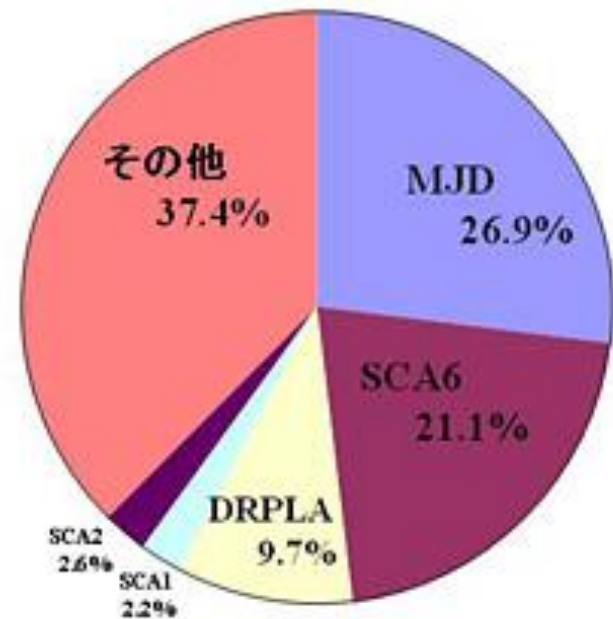
Naito, 1990 / Neurogenetics (1998) 2:1–17

わが国における脊髄小脳変性症の疫学



- sporadic SCD
- AD-SCD
- AR-SCD
- other-SCD
- SSP

遺伝性 SCD (n= 2,823)



- MJD
- SCA6
- DRPLA
- SCA1
- SCA2
- n_ADOthers

MR findings, late adult onset

- Atrophy the cerebellum, brainstem and superior cerebellar peduncle, in particular pontine tegmentum
- May be more appropriate to use the term “just small in size or hypoplastic” rather than atrophic
- Diffuse hyperintensity in the cerebral white matter on FLAIR and T2-weighted images in patients of long duration
- Hypointense area in the pontine base on T1-weighted image

MR findings in children

- Cerebellar atrophy, in early stage
- Brainstem and cerebral atrophy in advanced phase
- May be more appropriate to use the term “just small in size or hypoplastic” rather than atrophic
- Hyperintense area in the cerebral white matter on FLAIR and T2-weighted images in later stage

MR findings, early adult-onset

- Intermediate; halfway between late adult onset and juvenile onset