MRI showing white matter change

Exclude acquired conditions
Round 1 investigations
(supplementary Table 1 online)

Consider an Adult onset Leukodystrophy

A Family History (table 1)
  Autosomal dominant
  Autosomal recessive
  X-Linked
  Maternal inheritance

B Ethnicity (table 1)

C Clinical History and examination (table 2)
  Neurological
  Ophthalmological
  Systemic features
  Skin

D MRI pattern (Table 2, Figure 1)
  Brain and CS spine +/- Gad

Biochemical tests
Round 2 investigations (Table 4)

Genetic tests
See text

If diagnosis still elusive consider brain biopsy