
**Demographic:** Male, 57 yrs, America North, General speciality

**Clinical features extracted from the case and entered into the query box:**
- rapidly progressive dementia
- difficulty with speech
- forgetful
- impaired attention
- behaviour changes
- seizure
- elevated very long chain fatty acids

**Synonyms used by Isabel for the above query:**
- Dementia
- Seizure
- Forgetfulness
- Elevated VLCFA
- Difficulty with speech
- Behaviour changes

STOP!

Before you read further you might want to construct your own:

- Complete differential diagnosis
- Final diagnosis

In the following section you will find:

- The differential diagnosis constructed by the MGH panel
- The final diagnosis of the case
- Which of the MGH panel differential diagnoses did Isabel contain
- Did Isabel suggest the Final diagnosis
**Differential Diagnoses considered by the MGH panel:**
Progressive white-matter disease or leukoencephalopathy.

**Final Diagnosis of the case according to NEJM:**
Adult cerebral form of X-linked adrenoleukodystrophy

**Differential Diagnoses of the case as given by Isabel:**
Adult cerebral form of X-linked adrenoleukodystrophy – under Metabolic Diseases heading

**Was the final diagnosis given by Isabel:**
Yes, under Metabolic Diseases heading
### Isabel Differential:

**- NERVOUS SYSTEM DISORDERS**  >> click to collapse

- CVA / Stroke  
- Intracranial Abscess  
- Multiple Sclerosis  
- Frontotemporal Dementia  
- Alzheimer’s Disease  
- Vascular Dementia  
- Cerebral Sinus Venous Thrombosis  
- Mitochondrial Encephalomyopathies  
- Human Prion Diseases  
  - Creutzfeldt-Jakob Disease  
  - Paraneoplastic Neurologic Syndrome  
- Neuroacanthocytosis Disorders  
- Brain Neoplasms  
- Transient Global Amnesia  
- Progressive Supranuclear Palsy  
- Megalencephalic Leukodystrophy  
- Fahr’s Disease

**- INFECTIONOUS DISEASES**  >> click to collapse

- Measles Infection  
- Tuberculosis  
- Arboviral Infections / Encephalitides  
- Fungal Meningitis  
- Progressive Multifocal Leuкоencephalopathy  
- Syphilis  
- Limbic Encephalitis  
- Toxoplasmosis  
- Toxic Leukoencephalopathy  
- Bartonella Infection

**- GASTROINTESTINAL DISORDERS**  << click to expand

- Celiac Disease

**METABOLIC DISEASES**

- Adrenoleukodystrophy  

*Why did this diagnosis come up?*

*We matched the terms: mental deterioration | cognitive decline | dementia | delirium | seizures | seizure | difficulty with speech | change in behavior | change in behavior | elevated vWF | rapid | rapidly | progress | progressive*

*Degree of match between query entered and Isabel database (Not clinical probability): 58%*

**- RESPIRATORY DISORDERS**  >> click to collapse

- Sleep Apnea  
- Sarcoidosis  
  - Löfgren’s Syndrome  
  - Neurosarcoidosis

**- TOXICOLOGY**  << click to expand

- Heavy Metal Intoxication

**- RHEUMATIC DISEASES**  >> click to collapse

- Giant Cell Arteritis  
- Sjogren’s Syndrome  
- Lymphomatoid Granulomatosis  
- Antiphospholipid Syndrome

**- HUMAN GENETICS**  << click to expand

- Megaloblastic Anemias  
- TTP