

Case 1 (January 8 2009): [N Engl J Med. 360\(2\):171-181](#)

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

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Isabel Differential:

- NERVOUS SYSTEM DISORDERS >> click to collapse	- INFECTIOUS DISEASES >> click to collapse
<ul style="list-style-type: none"> CVA / Stroke ? RD Intracranial Abscess ? RD Multiple Sclerosis ? RD <ul style="list-style-type: none"> Frontotemporal Dementia ? RD Alzheimer's Disease ? RD Vascular Dementia ? RD Cerebral Sinus Venous Thrombosis ? RD Mitochondrial Encephalomyopathies ? RD Human Prion Diseases ? RD <ul style="list-style-type: none"> Creutzfeldt-Jakob Disease ? RD Paraneoplastic Neurologic Syndrome ? RD Neuroacanthocytosis Disorders ? RD Brain Neoplasms ? RD <ul style="list-style-type: none"> Transient Global Amnesia ? RD Progressive Supranuclear Palsy ? RD Megalencephalic Leukodystrophy ? RD Fahr's Disease ? RD 	<ul style="list-style-type: none"> Measles Infection ? RD Tuberculosis ? RD Arboviral Infections / Encephalitides ? RD Fungal Meningitis ? RD <ul style="list-style-type: none"> Progressive Multifocal Leucoencephalopathy ? RD Syphilis ? RD <ul style="list-style-type: none"> Limbic Encephalitis ? RD Toxoplasmosis ? RD <ul style="list-style-type: none"> Toxic Leukoencephalopathy ? RD Bartonella Infection ? RD
+ GASTROINTESTINAL DISORDERS << click to expand	+ GASTROINTESTINAL DISORDERS << click to expand
	<ul style="list-style-type: none"> Celiac Disease ? RD
- RESPIRATORY DISORDERS >> click to collapse	METABOLIC DISEASES
<ul style="list-style-type: none"> Sleep Apnea ? RD Sarcoidosis ? RD <ul style="list-style-type: none"> Lofgren' Syndrome Neurosarcoidosis 	<ul style="list-style-type: none"> Adrenoleukodystrophy ⚡ <input type="radio"/> RD <div style="border: 1px solid black; background-color: #ffff00; padding: 5px; margin-top: 10px;"> <p>Why did this diagnosis come up ?</p> <p>We matched the terms: mental deterioration cognitive decline dementia demential seizures seizure difficulty with speech change in behaviour change in behavior elevated vlcfα rapid rapidly progress progressive</p> <p>Degree of match between query entered and isabel database (Not clinical probability): 58%</p> <ul style="list-style-type: none"> Adrenomyeloleukodystrophy ⚡ Adrenomyeloneuropathy ⚡ X-linked Adult Cerebral Adrenoleukodystrophy ⚡ </div>
+ TOXICOLOGY << click to expand	- HEMATOLOGY >> click to collapse
<ul style="list-style-type: none"> Heavy Metal Intoxication ? RD 	<ul style="list-style-type: none"> Megaloblastic Anemias ? RD TTP ? RD
- RHEUMATIC DISEASES >> click to collapse	
<ul style="list-style-type: none"> Giant Cell Arteritis ? RD Sjogren's Syndrome ? RD Lymphomatoid Granulomatosis ? RD Antiphospholipid Syndrome ? RD 	
+ HUMAN GENETICS << click to expand	