



Case 25 (August 14 2008): [N Engl J Med. 359\(7\):736-747](#)

Demographic: Male, 43 years, America North, General speciality

Clinical features extracted from the case and entered into the query box:

insomnia
fatigue
joint pains
cold intolerance
anxiety
Diabetes Insipidus
weakness in proximal thighs
somnolence
ataxia
depressed mood
dysarthria
exophthalmos
bilateral hydronephrosis
muscle pains
gait imbalance

Synonyms used by Isabel for the above query:

Diabetes Insipidus
Insomnia
Fatigue
Joint pains
Cold intolerance
Anxiety
Somnolence
Ataxia
Depressed mood
Dysarthria
Exophthalmos
Muscle pains

STOP !

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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:

Sarcoidosis
Systemic histiocytosis

Final Diagnosis of the case according to NEJM:

Erdheim-Chester Disease

Differential Diagnoses of the case as given by Isabel:

Sarcoidosis – under Respiratory disorders heading
Erdheim-Chester Disease – under Neoplastic diseases heading

Was the final diagnosis given by Isabel:

Yes, Erdheim-Chester Disease under Neoplastic diseases heading

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Isabel Differential:

<p>+ NERVOUS SYSTEM DISORDERS << click to expand</p> <p><input type="checkbox"/> Migraine ? RD</p> <p><input type="checkbox"/> CVA / Stroke ? RD</p>	<p>+ INFECTIOUS DISEASES << click to expand</p> <p><input type="checkbox"/> Measles Infection ? RD</p> <p><input type="checkbox"/> Bacterial Meningitis ? RD</p> <p><input type="checkbox"/> Viral Meningoencephalitis ? RD</p>
<p>+ ENDOCRINE SYSTEM << click to expand</p> <p><input type="checkbox"/> Hypothyroidism ? RD</p>	<p>- RESPIRATORY DISORDERS >> click to collapse</p>
<p>+ GASTROINTESTINAL DISORDERS << click to expand</p> <p><input type="checkbox"/> Food Poisoning ? RD</p>	<p><input type="checkbox"/> Atypical Pneumonia ? RD</p> <p><input type="checkbox"/> Lung Neoplasms ? RD</p> <p><input type="checkbox"/> Sarcoidosis ? RD</p>
<p>+ RHEUMATIC DISEASES << click to expand</p>	<p>Why did this diagnosis come up ? ✖</p> <p>We matched the terms: diabetes insipidus di dysarthria daytime hypersomnolence depressed mood malaise fatigue arthralgias arthralgia weak weakness proxim proximal bilater bilateral</p> <hr/> <p>Degree of match between query entered and isabel database (Not clinical probability): 46%</p>
<p>- NEOPLASTIC DISEASES >> click to collapse</p> <p>Erdheim-Chester Disease ? RD</p>	<p><input type="checkbox"/> Sleep Apnea ? RD</p>
<p>Why did this diagnosis come up ? ✖</p> <p>We matched the terms: ataxia ataxy diabetes insipidus di dysarthria daytime hypersomnolence depressed mood joint pains joint pain proptosis exophthalmos weak weakness bilater bilateral gait imbal imbalance</p> <hr/> <p>Degree of match between query entered and isabel database (Not clinical probability): 32%</p>	<p>NUTRITIONAL DISORDERS</p> <p><input type="checkbox"/> Vit C Deficiency ? RD</p>
<p>Langerhans Cell Histiocytosis Class 1 ? RD</p>	<p>+ TOXICOLOGY << click to expand</p>
<p>+ OPHTHALMOLOGY << click to expand</p>	<p>+ CARDIAC DISORDERS << click to expand</p>
<p>+ NEUROMUSCULAR DISORDERS << click to expand</p>	<p>+ PSYCHOLOGIC DISORDERS << click to expand</p>
<p>+ METABOLIC DISEASES << click to expand</p>	<p>+ NEPHROLOGY << click to expand</p>
<p>+ ALLERGIC DISORDERS << click to expand</p>	<p>+ DERMATOLOGY << click to expand</p>
<p>+ LIVER DISORDERS << click to expand</p>	<p>+ ENVIRONMENTAL HEALTH HAZARDS << click to expand</p>