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**Case 32 (October 15 2009):** [N Engl J Med. 361\(16\):1585-1593](#)

**Demographic:** Male, 27yrs, America North, General speciality

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

increased shortness of breath  
headaches  
weight gain  
oculocutaneous albinism  
tachycardia  
digital clubbing

**Synonyms used by Isabel PRO for the above query:**

digital clubbing  
headaches  
tachycardia  
weight gain  
oculocutaneous albinism

**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

**Final Diagnosis of the case according to NEJM:**

Hermansky-Pudlak Syndrome

**Was the final diagnosis given by Isabel PRO:**

Yes, Hermansky-Pudlak Syndrome in Metabolic diseases.

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### Isabel PRO Final Diagnosis:

**ENTER CLINICAL FEATURES**      **Synonyms**      **DEGREE OF MATCH**      **Your Customized View**

**age**      young adult (16 - 30 years) ▾  
**gender**      male ▾  
**pregnancy**      (- not specified -) ▾  
**region**      America North ▾ ⓘ  
**speciality**      General ▾

ENTER QUERY TERMS ON SEPARATE LINES: ⓘ

increased shortness of breath
headaches
weight gain
oculocutaneous albinism
tachycardia
digital clubbing

+ Add a Clinical Feature

**Get Checklist »**

[Help](#)      [Search Again](#)

Isabel PRO is not meant to replace your clinical judgment.

**45 DIAGNOSES- DEGREE OF MATCH**

<input checked="" type="checkbox"/> Hypersensitivity Pneumonitis	? RD
<input checked="" type="checkbox"/> Diseases of High Altitudes	? RD
<input checked="" type="checkbox"/> Hepatocellular Failure	? RD
<input type="checkbox"/> Hermansky-Pudlak Syndrome	RD

**Why did this diagnosis come up ?** ⓘ  
We matched the terms: albinism | albino | breath

**Degree of match between query entered and Isabel database (Not clinical probability): 22%**

<input checked="" type="checkbox"/> Drug Overdose/Poisoning	? RD
<input checked="" type="checkbox"/> Environmental (Work) Exposure	? RD

1 2 3 4 5

**14 Body Systems**

- HEMATOLOGY DISORDERS
- ENDOCRINE DISORDERS
- GASTROINTESTINAL DISORDERS
- LIVER DISORDERS
- INFECTIOUS DISEASES
- NEPHROLOGY DISORDERS
- NUTRITIONAL DISORDERS
- SHOCK STATES
- PSYCHOLOGIC DISORDERS
- TOXICOLOGY DISORDERS
- ENVIRONMENTAL HEALTH DISORDERS
- METABOLIC DISEASES

**SHOW ALL DIAGNOSES**