

Case 27 (August 29 2013): [N Engl J Med. 369\(9\):853-863](#)

Demographic: Male, 6.5 months old, North America

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

fever
thrombocytopenia
easy bruising
petechial rash
splenomegaly

Synonyms used by Isabel for the above query:

fever
thrombocytopenia
easy bruising
petechial rash
splenomegaly

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:

ITP
Common Variable Immunodeficiency (CVID)

Final Diagnosis of the case according to NEJM:

Autoimmune Lymphoproliferative Syndrome (ALPS) due to a germline FAS mutation

Differential Diagnoses of the case as given by Isabel:

ITP under Hematology
Common Variable Immunodeficiency (CVID) under Immunology

Was the final diagnosis given by Isabel:

Yes, Autoimmune Lymphoproliferative Syndrome (ALPS) under Immunology

Isabel Checklist Differential:

enter clinical features | synonyms

age*

gender female male

Refine search:

travel history:

show me:

diagnoses

causative drugs

bioterrorist agents

Enter clinical features, no negatives, no numbers:

- fever
- thrombocytopenia
- easy bruising
- petechial rash
- splenomegaly

+ add a clinical feature

get checklist ➔

[clear search](#)

Isabel is not meant to replace your clinical judgment.

diagnoses | drugs

show 10 | show all | don't miss

Autoimmune Lymphoproliferative Syndrome IMMUNO

Why did this diagnosis come up ?

We matched the terms: fever | fevers | feverish | feverishness | thrombocytopenia | thrombocytopenic | easy bruising | petechial rash | petechiae | petechia | petechial spots | petechial hemorrhage | petechial haemorrhage | petechial spots on soft palate | petechial spots on palate | petechial spots buccal mucosa | splenomegaly | splenomegaly

Degree of match between query entered and Isabel database (Not clinical probability): 90%

ITP	👍 👎	? HEMAT
Rocky Mountain Spotted Fever	👍 👎	? INFEC
Vit C Deficiency	👍 👎	? NUTRI
Brucellosis	👍 👎	? INFEC
+ Liver Neoplasms	👍 👎	? NEOPL
Measles	👍 👎	? INFEC
Meningococcal Disease	👍 👎	? INFEC

Click diagnosis for evidence-based content.

feedback: