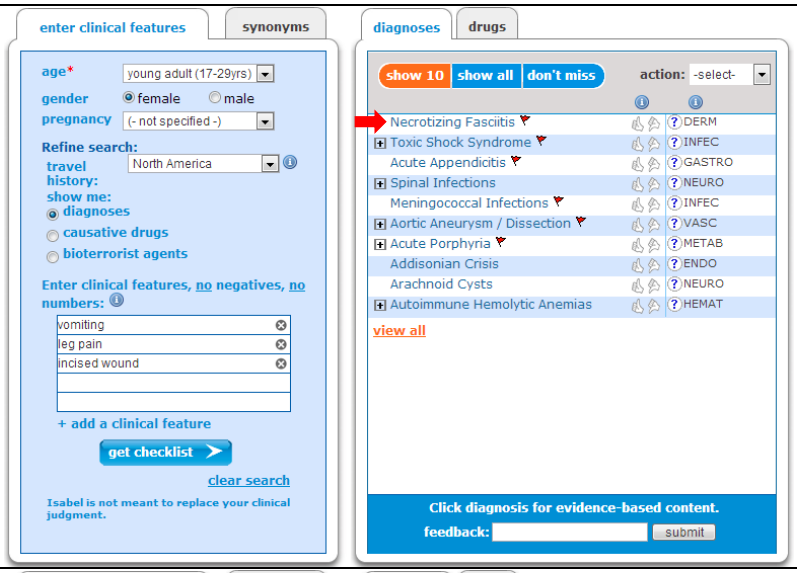
Rare/Uncommon Diseases:

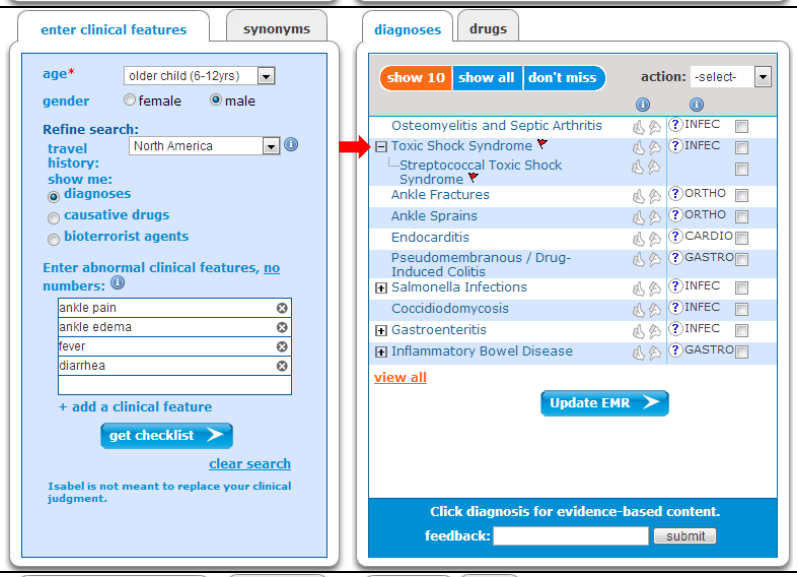
Dx: Uncommon; Grade: Simple; Symptoms: Typical

A 24 year old female presented in the ED with a deep cut in her calf as the result of a homemade zipline accident where she fell into a brackish river. The cut was cleaned and closed with 22 stitches. The patient was discharged with pain medication instructions. The following day she returned with intense leg pain and vomiting.



Dx: Uncommon; Grade: Simple; Symptoms: Typical

A 10 year old male presented in the ED with ankle pain and ankle edema as the result of a basketball injury. The ankle was wrapped and the patient discharged with pain medication instructions. The following day he returned with fever and diarrhea.



Dx: Uncommon; Grade: Complex; Symptoms: Atypical

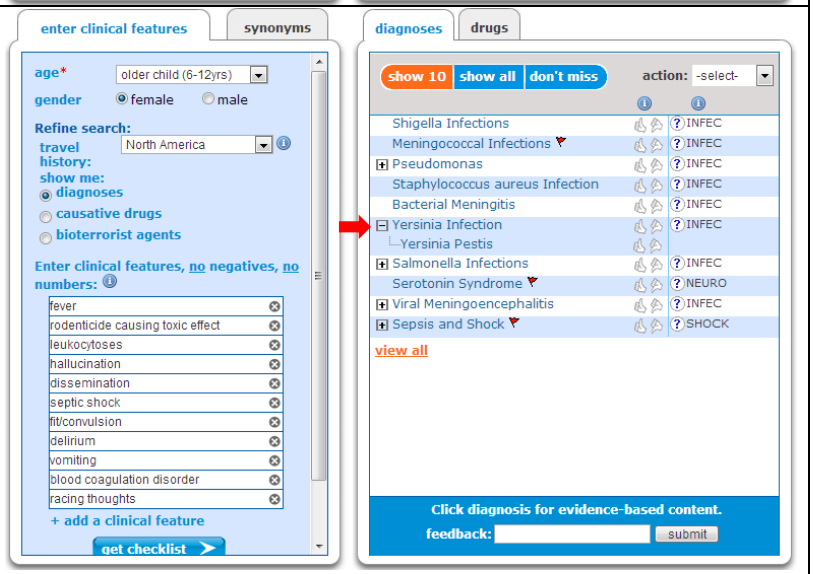
Sierra Jane Downing had come upon a half-eaten squirrel while on a picnic with her family on Aug.19 in Pagosa Springs.

Sierra Jane woke up with a fever and was vomiting, and by 9 p.m. her father found her lying on the bathroom floor. When he picked her up to bring her back to bed, the girl threw up again and then had a seizure.

Sierra Jane's clear chest X-ray and only slightly elevated white blood cell. They managed to bring Sierra Jane's temperature down from 107 to 103 degrees. By 6:30 p.m. Saturday, Sierra Jane's condition had worsened and she was transferred to pediatric ICU for septic shock. Doctors initially thought that Sierra Jane's altered mental state -- which included delirium and hallucinations -- was a result of a dose of the antibiotic ceftriaxone.

Dr. Jennifer Snow, the pediatric critical care specialist whose quick thinking proved so important, ran some blood tests on Sierra Jane, as the girl was in disseminated intravascular coagulopathy, meaning her blood would not clot.

"Mom told us the story of the exposure to the dead squirrel, and exposure to mouse droppings in chicken coops, and exposure to a dead skunk," Snow told ABC News. Snow did an immediate literature search, and found a report of a 16-year-old with fulminant septic shock in the chest whose cause of death was listed as bubonic plague.

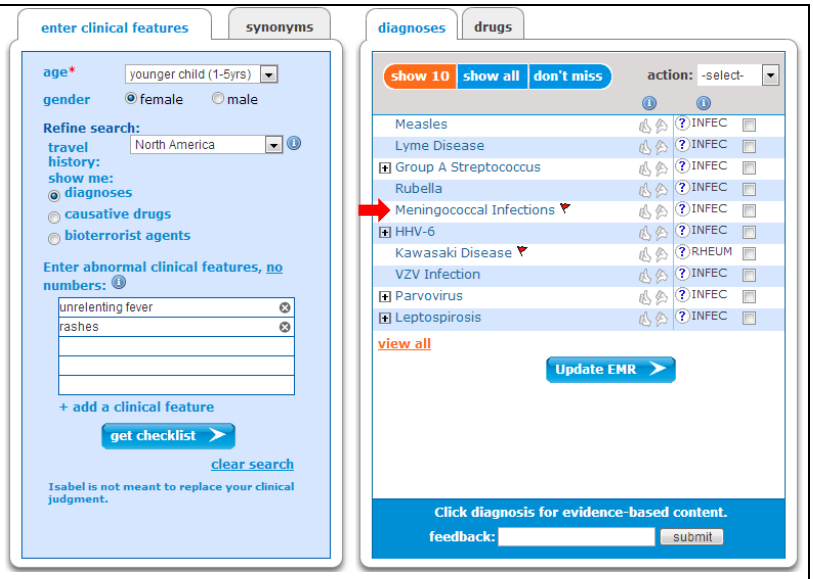


Dx: Uncommon; Grade: Simple; Symptoms: Typical

Georgia, a two-year-old girl was diagnosed with swine flu. She was given Tamiflu in order to control her fever, but her condition kept on deteriorating.

Her parents were of the view that there are chances that their elder daughter, Charlie's, swine flu condition would have added to the confusion. After coming across the confusion, the Ambulance Service apologized to Georgia's parents. The paramedic who treated her was of the view that she was misdiagnosed. She had rashes, which is one of the main symptoms of meningitis. The paramedic further affirmed that she diagnosed Georgia with swine flu after looking at her history.

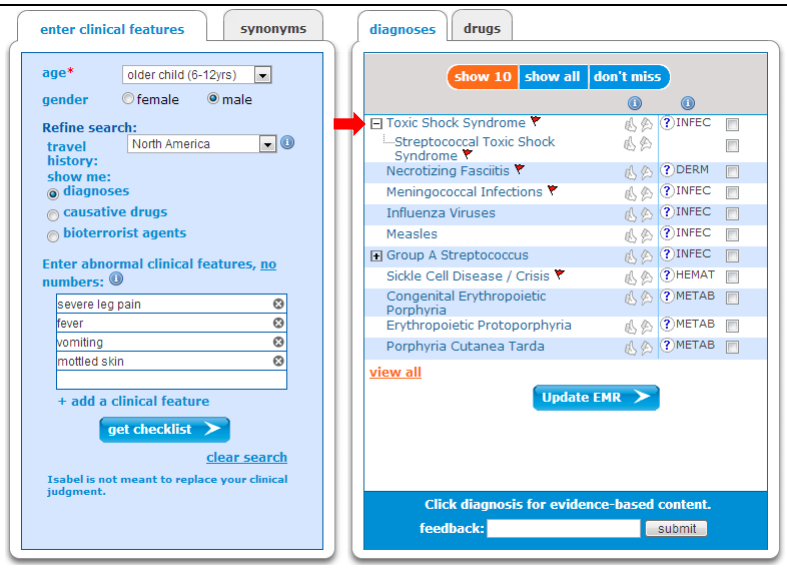
Georgia's medical report at the University Hospital revealed that she was suffering from blood poisoning that was caused by meningitis.



Dx: Uncommon; Grade: Simple; Symptoms: Typical

Rory, a 5-foot-9, 169-pound sixth grader from Queens, cut his arm while diving for a basketball one spring afternoon in the school gym. He began vomiting after midnight. By the time he saw the family pediatrician later that day, he was running a high fever and suffering severe leg pain, and his skin was not returning to its normal color quickly when pressed with a finger (mottled skin). The vomiting and fever suggested a stomach bug to the pediatrician, who sent him to the emergency room at NYU Langone for fluids. Taken together, those signs suggested that he could be in the grip of something more worrisome than a stomach virus. A doctor in the emergency room thought he looked better after some intravenous liquids. He went home with an antinausea drug.

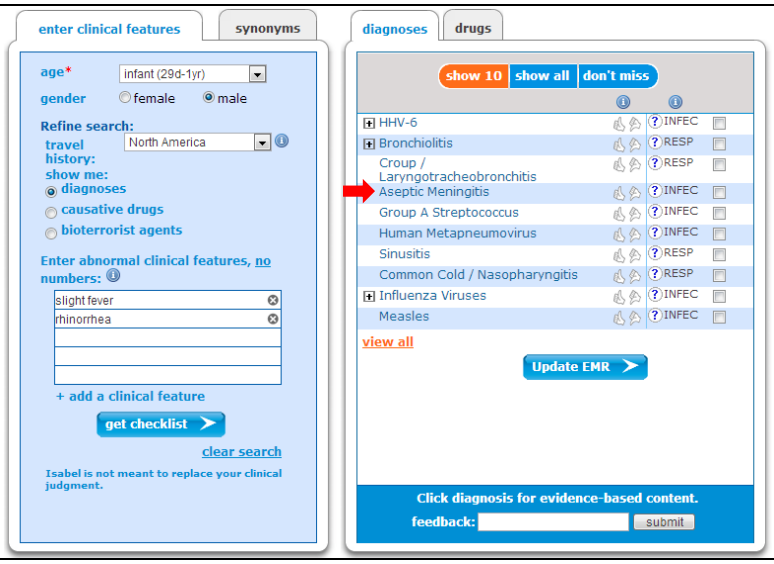
Rory's body, however, was fighting an invader. His immune system was on the verge of a runaway, self-immolating response to the infection — a cascade of destructive processes known generally as sepsis.



Dx: Uncommon; Grade: Complex; Symptoms: Atypical

Daniel Pranson, MD, a pediatrician in Chicago with a reputation as a top-notch diagnostician, described an instance of finding the less obvious diagnosis. A toddler presented with slight fever and rhinorrhea, and he suggested a decongestant. Two days later, the mother returned saying, "He's not acting like himself." Dr. Pranson accurately suspected meningitis. What made Dr. Pranson jump to the zebra instead of the horse? "I'm no genius, and I wasn't the class valedictorian," says Dr. Pranson. "I try to stay current with the literature, but more important, I look at subtleties. And I regard a mother's return visit as a red flag, which I trust. Parents can sense when something isn't right with their child. This motivated me to look more carefully under the surface presentation."

[Are You a Great Diagnostician? - MedScape]

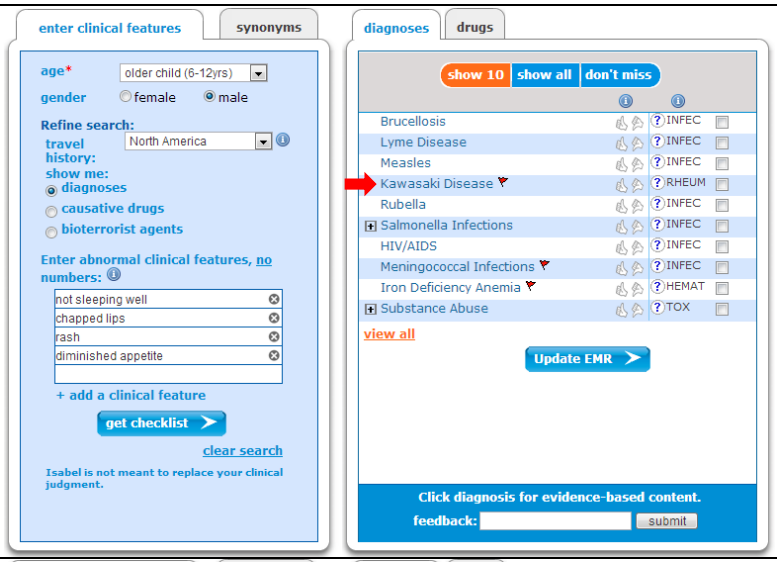


Dx: Uncommon; Grade: Complex; Symptoms: Typical

Grey's Anatomy – Kawasaki Disease segment of the show which aired Thursday, March 28, 2013.

The preview contained the symptom information – not sleeping well, chapped lips, rash, diminished appetite - Grey's Anatomy Sneak Preview.

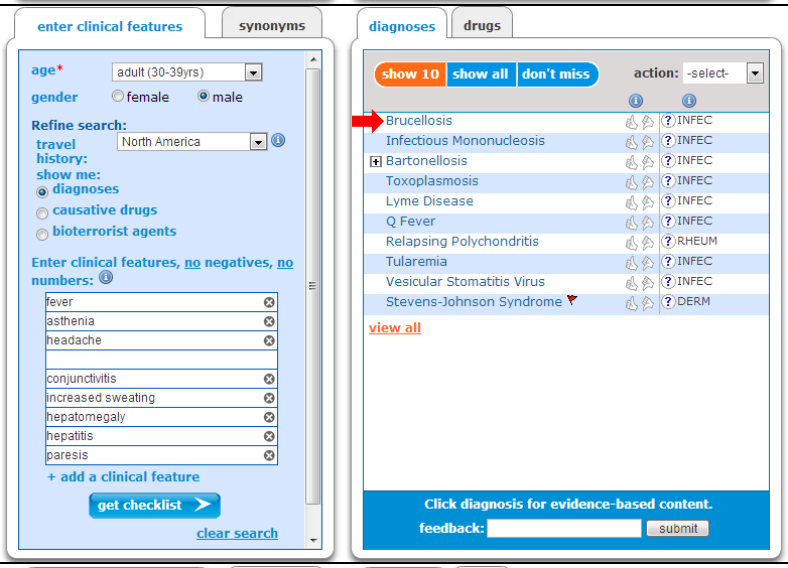
Kawasaki Disease is commonly not picked up and if not treated quickly leads to aneurysms throughout the child's life.



Dx: Uncommon; Grade: Complex; Symptoms: Atypical

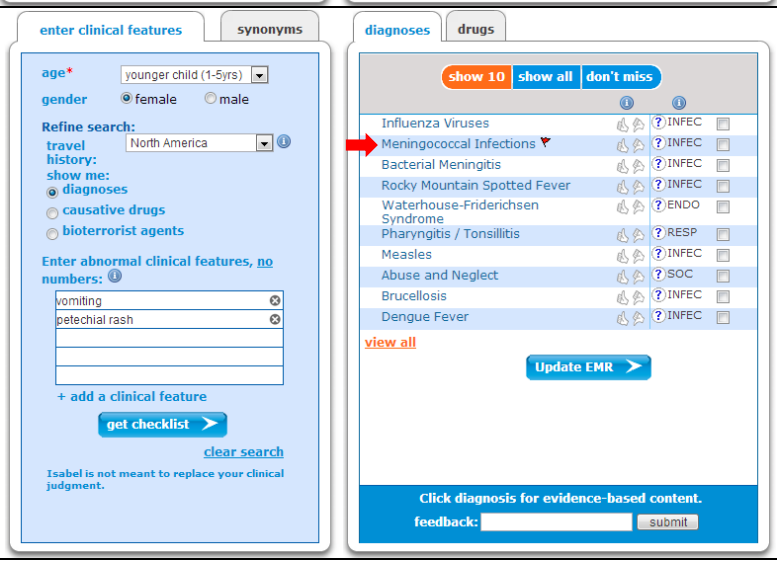
Resident Rounding – "at morning report we had a male patient, 33 years old, with headache. As the story evolved, the patient also had fever, hepatomegaly and conjunctivitis. Later added sweating and weakness."

The diagnosis obtained by liver biopsy and lots of exclusions was Brucellosis. This was not on our differential diagnosis. When I typed these terms into Isabel (afterwards) the first diagnosis on the list was Brucellosis! I will take this search to morning report tomorrow to show the residents and two of our educational leaders.



Dx: Uncommon; Grade: Simple; Symptoms: Typical

On February 25, 2012, Kelsey was a happy, healthy, and popular 5 year old girl who fell ill with vomiting and a small pinprick rash on her abdomen and left upper leg. The doctor who first saw Kelsey said it would be better to take her home and go to hospital the next day if she was still ill. Giving evidence the doctor stated: "In hindsight, yes, I should have referred her to hospital, but I thought I was dealing with viral gastroenteritis and I couldn't imagine what was behind her very non-specific symptoms. My main concern was the vomiting. If there had been more signs pointing to meningitis I would have drawn that conclusion, I would expect signs to be much more obvious." The second doctor to examine Kelsey said she did think about meningitis, but discredited that at the time.
She stated: "The rash didn't seem to have progressed. If things are stable and no worse you are reassured. The rash was sort of non-specific and didn't look like anything I had expected a meningitis rash to look like." The girl died from meningitis just hours after the two doctors had misdiagnosed her symptoms as a stomach bug.



Dx: Uncommon; Grade: Complex; Symptoms: Typical

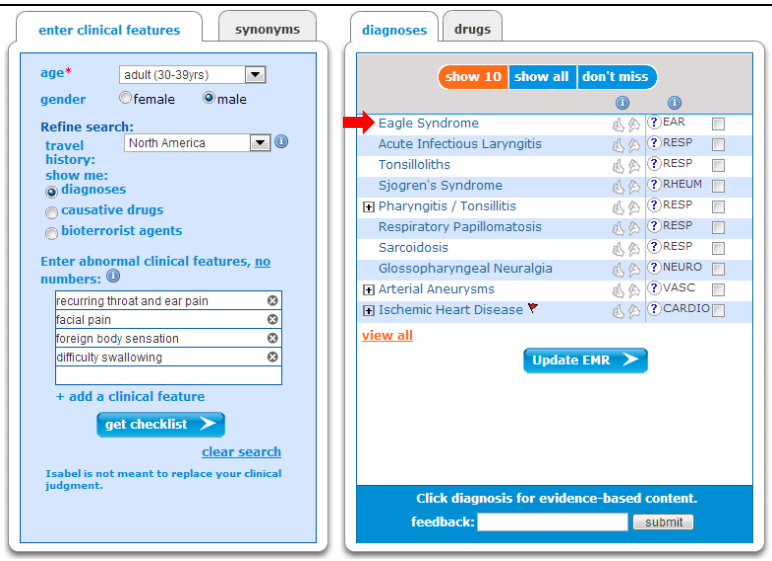
Carl Holt, 34, was subjected to two years of GP and hospital appointments suffering constant throat pain after he fell ill on a family holiday in July 2010, visiting doctors 150 TIMES. Despite hospital appointments and visits to his doctor up to SIX times a month, medics could not work out what was wrong with him.

But after researching his symptoms on the internet and watching videos on Youtube, he found out about a rare condition called Eagle Syndrome.

Remarkably, despite telling doctors about the disease, Carl was never tested for it at three separate GP practices or the University Hospital of North Staffordshire.

Carl then decided to pay for private care at Salford Hospital in Greater Manchester in October last year and was finally diagnosed with the rare medical condition within MINUTES of his first visit.

Symptoms of Eagle Syndrome include throat and ear pain as a result of small bones to the rear of the throat becoming calcified, or hardened. Sufferers often have the feeling something is stuck in their throat and have difficult swallowing as well as facial pains.



Dx: Uncommon; Grade: Complex; Symptoms: Atypical

I became ill and it took seventeen weeks of investigations by five hospitals to make a diagnosis. I am told that the variable manner in which Giant Cell Arteritis (GCA) develops symptoms can make it indistinguishable from many other diseases and thus poses a diagnostic problem for doctors.

Is GCA hard to diagnose though? I Googled 'the world's best medical diagnostician'. The result took me to a software package called Isabel. I used this software to investigate my symptoms as they had developed over several months.

WEEK ONE: Whilst on holiday in France, the symptoms of my illness were; lower abdominal discomfort, constant headache, weight loss, nausea, tired and unwell. I had three symptoms that I did not describe but were present they were dry cough, scalp irritation and fever.

WEEK FOUR: At week four I had the following additional symptoms; chest tightness, night sweats, neck pain, sore throat and hoarse voice.

WEEK SIX: I had acquired further symptoms of deep dry cough, sharp chest pain and the need to take shallow breaths to avoid coughing.

WEEK SEVEN: With further symptoms of jaw and face ache and pain in my ears.

The cost to the NHS of my stays in hospital and diagnostic procedures was in excess of $31,000 US $$. The use of Isabel in the early stages of my illness would have saved 95% of these costs and prevented some irreversible damage to my arteries.