

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.

enter clinical features

synonyms

age*

young adult (17-29yrs)

gender

☒ female ☐ male

pregnancy

(- not specified -)

Refine search:

travel

North America

history:

show me:

☒ diagnoses

☐ causative drugs

☐ bioterrorist agents

Enter clinical features, no negatives, no numbers:

vomiting

leg pain

incised wound

+ 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 action: -select-

Necrotizing Fasciitis

Toxic Shock Syndrome

Acute Appendicitis

Spinal Infections

Meningococcal Infections

Aortic Aneurysm / Dissection

Acute Porphyria

Addisonian Crisis

Arachnoid Cysts

Autoimmune Hemolytic Anemias

view all

Click diagnosis for evidence-based content.

feedback: submit

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.

enter clinical features

synonyms

age*

older child (6-12yrs)

gender

☐ female ☒ male

Refine search:

travel

North America

history:

show me:

☒ diagnoses

☐ causative drugs

☐ bioterrorist agents

Enter abnormal clinical features, no numbers:

ankle pain

ankle edema

fever

diarrhea

+ 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 action: -select-

Osteomyelitis and Septic Arthritis

Toxic Shock Syndrome

Streptococcal Toxic Shock Syndrome

Ankle Fractures

Ankle Sprains

Endocarditis

Pseudomembranous / Drug-Induced Colitis

Salmonella Infections

Coccidioidomycosis

Gastroenteritis

Inflammatory Bowel Disease

view all

Update EMR

Click diagnosis for evidence-based content.

feedback: submit

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.

enter clinical features

synonyms

age*

older child (6-12yrs)

gender

☒ female ☐ male

Refine search:

travel

North America

history:

show me:

☒ diagnoses

☐ causative drugs

☐ bioterrorist agents

Enter clinical features, no negatives, no numbers:

fever

rodenticide causing toxic effect

leukocytoses

hallucination

dissemination

septic shock

fit/convulsion

delirium

vomiting

blood coagulation disorder

racing thoughts

+ add a clinical feature

get checklist

diagnoses

drugs

show 10 show all don't miss action: -select-

Shigella Infections

Meningococcal Infections

Pseudomonas

Staphylococcus aureus Infection

Bacterial Meningitis

Yersinia Infection

Yersinia Pestis

Salmonella Infections

Serotonin Syndrome

Viral Meningoencephalitis

Sepsis and Shock

view all

Click diagnosis for evidence-based content.

feedback: submit

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.

enter clinical features

synonyms

age*
younger child (1-5yrs)

gender
☒ female ☐ male

Refine search:
travel history:
North America

show me:
☒ diagnoses
☐ causative drugs
☐ bioterrorist agents

Enter abnormal clinical features, no numbers:
unrelenting fever
rashes

+ 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

action: -select-

Measles

?

INFE

Lyme Disease

?

INFE

Group A Streptococcus

?

INFE

Rubella

?

INFE

Meningococcal Infections

?

INFE

HHV-6

?

INFE

Kawasaki Disease

?

RHEUM

VZV Infection

?

INFE

Parvovirus

?

INFE

Leptospirosis

?

INFE

view all

Update EMR

Click diagnosis for evidence-based content.

feedback: submit

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.

enter clinical features

synonyms

age*
older child (6-12yrs)

gender
☐ female ☒ male

Refine search:
travel history:
North America

show me:
☒ diagnoses
☐ causative drugs
☐ bioterrorist agents

Enter abnormal clinical features, no numbers:
severe leg pain
fever
vomiting
mottled skin

+ 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

action: -select-

Toxic Shock Syndrome

?

INFE

Streptococcal Toxic Shock Syndrome

?

INFE

Necrotizing Fasciitis

?

DERM

Meningococcal Infections

?

INFE

Influenza Viruses

?

INFE

Measles

?

INFE

Group A Streptococcus

?

INFE

Sickle Cell Disease / Crisis

?

HEMAT

Congenital Erythropoietic Porphyria

?

METAB

Erythropoietic Protoporphyria

?

METAB

Porphyria Cutanea Tarda

?

METAB

view all

Update EMR

Click diagnosis for evidence-based content.

feedback: submit

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](#)

enter clinical features

synonyms

age*
infant (29d-1yr)

gender
☐ female ☒ male

Refine search:
travel history:
North America

show me:
☒ diagnoses
☐ causative drugs
☐ bioterrorist agents

Enter abnormal clinical features, no numbers:
slight fever
rhinorrhea

+ 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

action: -select-

HHV-6

?

INFE

Bronchiolitis

?

RESP

Croup / Laryngotracheobronchitis

?

RESP

Aseptic Meningitis

?

INFE

Group A Streptococcus

?

INFE

Human Metapneumovirus

?

INFE

Sinusitis

?

RESP

Common Cold / Nasopharyngitis

?

RESP

Influenza Viruses

?

INFE

Measles

?

INFE

view all

Update EMR

Click diagnosis for evidence-based content.

feedback: submit

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.

enter clinical features synonyms

age* older child (6-12yrs) ▼

gender ☐ female ☒ male

Refine search:
travel history: North America ▼ ⓘ

show me:
☒ diagnoses
☐ causative drugs
☐ bioterrorist agents

Enter abnormal clinical features, no numbers: ⓘ

not sleeping well ⓘ
chapped lips ⓘ
rash ⓘ
diminished appetite ⓘ

+ 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

Brucellosis	?	INFE	
Lyme Disease	?	INFE	
Measles	?	INFE	
Kawasaki Disease ▼	?	RHEU	
Rubella	?	INFE	
Salmonella Infections	?	INFE	
HIV/AIDS	?	INFE	
Meningococcal Infections ▼	?	INFE	
Iron Deficiency Anemia ▼	?	HEMAT	
Substance Abuse	?	TOX	

view all

Update EMR >

Click diagnosis for evidence-based content.

feedback: submit

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.

enter clinical features synonyms

age* adult (30-39yrs) ▼

gender ☐ female ☒ male

Refine search:
travel history: North America ▼ ⓘ

show me:
☒ diagnoses
☐ causative drugs
☐ bioterrorist agents

Enter clinical features, no negatives, no numbers: ⓘ

fever ⓘ
asthenia ⓘ
headache ⓘ

conjunctivitis ⓘ
increased sweating ⓘ
hepatomegaly ⓘ
hepatitis ⓘ
paresis ⓘ

+ 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 action: -select- ▼

Brucellosis	?	INFE	
Infectious Mononucleosis	?	INFE	
Bartonellosis	?	INFE	
Toxoplasmosis	?	INFE	
Lyme Disease	?	INFE	
Q Fever	?	INFE	
Relapsing Polychondritis	?	RHEU	
Tularemia	?	INFE	
Vesicular Stomatitis Virus	?	INFE	
Stevens-Johnson Syndrome ▼	?	DERM	

view all

Click diagnosis for evidence-based content.

feedback: submit

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.

enter clinical features synonyms

age* younger child (1-5yrs) ▼

gender ☒ female ☐ male

Refine search:
travel history: North America ▼ ⓘ

show me:
☒ diagnoses
☐ causative drugs
☐ bioterrorist agents

Enter abnormal clinical features, no numbers: ⓘ

vomiting ⓘ
petechial rash ⓘ

+ 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

Influenza Viruses	?	INFE	
Meningococcal Infections ▼	?	INFE	
Bacterial Meningitis	?	INFE	
Rocky Mountain Spotted Fever	?	INFE	
Waterhouse-Friderichsen Syndrome	?	ENDO	
Pharyngitis / Tonsillitis	?	RESP	
Measles	?	INFE	
Abuse and Neglect	?	SOC	
Brucellosis	?	INFE	
Dengue Fever	?	INFE	

view all

Update EMR >

Click diagnosis for evidence-based content.

feedback: submit

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.

enter clinical features

synonyms

age*

adult (30-39yrs)

gender

female

male

Refine search:

travel history:

North America

show me:

diagnoses

causative drugs

bioterrorist agents

Enter abnormal clinical features, no numbers:

recurring throat and ear pain

facial pain

foreign body sensation

difficulty swallowing

+ 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

Eagle Syndrome

Acute Infectious Laryngitis

Tonsilloliths

Sjogren's Syndrome

Pharyngitis / Tonsillitis

Respiratory Papillomatosis

Sarcoidosis

Glossopharyngeal Neuralgia

Arterial Aneurysms

Ischemic Heart Disease

view all

Update EMR

Click diagnosis for evidence-based content.

feedback:

submit

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.

enter clinical features

synonyms

travel history:

North America

show me:

diagnoses

causative drugs

bioterrorist agents

Enter abnormal clinical features, no numbers:

lower abdominal discomfort

constant headache

weight loss

dry cough

tired

scalp irritation

fever

chest tightness

night sweats

neck pain

sore throat

hoarse voice

deep dry cough

sharp chest pain

shallow breathing

jaw ache

face ache

ear pain

diagnoses

drugs

show 10

show all

don't miss

Giant Cell Arteritis

Sarcoidosis

Non-Hodgkin Lymphoma

Influenza

CMV Infections

Sjogren's Syndrome

Babesiosis

Tularemia

Atypical Pneumonia

Lyme Disease

view all

Update EMR

Click diagnosis for evidence-based content.

feedback:

submit