Case 2
Acknowledgements: The author would like to thank Drs. Joel Fish, MSc, MD, FRCS and Manuel Gomez, MD, Sunnybrook & Women’s College Health Sciences Centre, for their contributions to this case.

Ms. H, a 35 year old woman involved in a house fire, arrived at the emergency department with vital signs absent. Her pupils were fixed and dilated, and black soot covered her face and neck. CPR was immediately initiated, and Ms. H was resuscitated. However, she was unresponsive to even painful stimuli. Her Glasgow coma score was 4.

Upon physical examination, her burn injury was minor - involving a total body surface area (TBSA) of 4%. She remained in a comatose state. Her blood gases revealed a pO2=75mmHg, pCO2=30mmHg, and pH 6.76 on a FiO2 of 100%.

An MRI of the brain taken 3 hours after the accident is shown below.

What is the Diagnosis?
Case 3

Acknowledgement
The author would like to thank Dr. Donald Rosenthal, M.D., McMaster University Medical Centre, for his contribution to this case.

An eleven-year-old girl presented to the dermatology clinic complaining of poor wound healing on her knees ever since she lacerated them from a minor injury at age seven. She was an otherwise healthy girl, with no significant past medical history. On review of systems, she admitted to being quite flexible in her joints and also pointed out small bumps on her wrists. Her family history was unremarkable.

On examination, actual scarring and cyst formation was seen bilaterally on the knees (Figure 3.1). The patient’s joints were indeed very flexible—she was able to touch her thumb flat to her forearm (Figure 3.2). She also demonstrated positive Gorlin’s sign, reaching her nose with her tongue. Increased elasticity of the skin, particularly over the joints was evident, and small lipoma-like lesions were found on flexor surfaces of both of her wrists. The rest of examination, including a thorough cardiovascular assessment, was unremarkable.

What is the likely Diagnosis?

Case 4

Acknowledgement
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A 37-year-old man presented complaining of intensely itching and burning eruption on his elbows lasting over three months. He was otherwise healthy, with no significant past medical history and unremarkable family history. He recalled that the lesions appeared shortly following his return from a fishing trip, where his diet consisted mostly of the caught fish, fried with wheat flour.

Physical examination revealed a young man in mild distress from his condition. He had erythematous papules, vesicles, and bullae symmetrically distributed on the extensor forearm and elbow surfaces, in grouped arrangement (Figure 4.1). The rest of skin examination as well as basic physical check-up were unremarkable.

What is the likely diagnosis?

What are the common associated conditions?

Figure 3.1. Photograph of patient’s knees

Figure 4.1. Photograph of lesions on patient’s extensor forearms and elbows

Figure 3.2. Patient’s joint flexibility
Case 1
A 22 year old man presents to you, his family physician, with recurrent chest pain. The previous week, he experienced a sudden onset of chest pain with palpitations while waiting for a friend to play squash at the local YMCA. His chest pain was accompanied by presyncope, nausea, shortness of breath, paresthesia in his extremities, hot flushes, and diaphoresis. He also had an overwhelming fear that he was going to die. An ambulance was called by the YMCA staff, and he was taken to the emergency department (ED). At the ED, an ECG, chest X-ray, and blood tests (CBC, glucose, CK) were ordered. All of the tests were normal. The ED doctor gave him 2mg of lorazepam and told him to follow up with his family physician. You perform a complete physical exam, and your only finding is some tenderness of the chest wall. You order an upper G.I. series, another ECG, and additional blood tests, including a test for H. pylori. You also refer him to a cardiologist who performs a stress test, echocardiogram, and 24 hour Holter monitor on your patient. All of the tests are normal.

During the 3 months your patient waited to see the cardiologist, he visited the ED on three separate occasions with complaints similar to his first visit. When you meet with your patient to discuss the test results and the cardiologist’s report, he has become reluctant to leave his apartment and increasingly depressed.

What is the Diagnosis?

And the diagnosis is . . .

Case 2
Diagnosis: Carbon monoxide (CO) poisoning
Smoke inhalation is a serious, potentially life-threatening injury that should be suspected in accidents involving fires or hot fumes in closed, confined spaces. Patients presenting with soot on the face, nose, or mouth should be examined for smoke inhalation injury. Those patients that give a clear history of being trapped in a closed space for more than two or three minutes have a high likelihood of sustaining a smoke inhalation injury and possibly carbon monoxide poisoning. In today’s homes, modern materials found in furnishings and building supplies may contribute to the products of combustion found in smoke.

For our patient, a blood carboxyhemoglobin (COHb) level was ordered immediately. As anticipated, the COHb level was elevated at 27%. Normal levels are <5% in non-smokers and <12% in smokers. Since CO is rapidly cleared from the Hb by breathing normal air or supplemental oxygen, the original COHb level in Ms. H was undoubtedly much higher. The half life of COHb is 20 minutes with hyperbaric oxygen therapy, 40 minutes after delivery of 100% supplemental oxygen, and 4 to 6 hours when breathing room air at atmospheric pressure.

Patients with COHb levels between 20-30% can present with symptoms of headache, dizziness, and confusion. Levels above 30% can lead to shock, coma, and death if the duration of exposure is long enough. Radiologically, MRI brain images may reveal symmetric abnormal signals in the globus pallidus of patients with CO poisoning. In serious cases, there may be a total loss of the normal brain convolutions.

References

Case 3
Diagnosis: Ehlers-Danlos Syndrome
The clinical features are consistent with the diagnosis of Ehlers-Danlos syndrome (EDS), a collagen defect of the extracellular matrix in various tissues. Eleven genetically, biochemically, and clinically diverse subtypes of EDS are defined at present, primarily by the extent to which skin, joints, and other tissues are

Laboratory findings generally include anemia and thrombocytopenia, hyponatremia, hyperkalemia, hypocalcemia, and metabolic acidosis. The anemia is characterized by negative direct and indirect Coombs tests and a smear consistent with mechanical destruction of erythrocytes in small vessels.

Early comprehensive management including dialysis therapy, aggressive management of hypertension, fresh frozen plasma transfusion, and nutritional support improves the outcome and decreases the mortality and morbidity in patients with HUS. Prognosis is generally quite good with less than 5% mortality. 85% of affected patients recover normal renal function.

About 5% of cases of E. coli 0157:H7 infection are complicated by the hemolytic-uremic syndrome (HUS), which is characterized by hemolytic anemia, thrombocytopenia, and acute renal failure. HUS typically has a prodrome of bloody diarrhea occurring five to seven days before onset of renal insufficiency. Colonoscopy is generally non-specific and shows hemorrhagic colitis. At the time of diagnosis, most children are extremely pale and very irritable.

Case 5
A 5 year old child was brought to the ED by her parents. The child had been playing in the dirt and was $(0.157:17)$ and similar strains of enterohemorrhagic E. coli produce high levels of Shiga toxins in the large intestine following infection. They cause direct mucosal damage, have a toxic effect on endothelial cells in the gut wall blood vessels, and once absorbed, create toxic effects on other vascular endothelia, such as the glomerular endothelial cells.

Of the many E. coli serotypes producing Shiga toxin, the 0157:H7 serotype is the most common in North America. Infection can occur in persons of all ages, although severe infection is most common in children and the elderly. Since this particular serotype has a bovine reservoir, outbreaks and cases of hemorrhagic colitis often occur after ingestion of undercooked beef or unpasteurized milk. Food or water contaminated with cow manure or raw ground beef can also transmit infection. The organism can also be transmitted among persons (especially among infants in diapers) by the fecal-oral route.

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E. coli O157:H7 and similar strains of enterohemorrhagic E. coli produce high levels of Shiga toxins in the large intestine following infection. They cause direct mucosal damage, have a toxic effect on endothelial cells in the gut wall blood vessels, and once absorbed, create toxic effects on other vascular endothelia, such as the glomerular endothelial cells.

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involved. The underlying molecular defect is believed to be due to mutations in the fibrillar collagen genes or enzymes that catalyse post-translational modification. The inheritance mode is autosomal dominant for most subtypes. The incidence may be 1 in 5000 births, although clinically mild cases might be under-reported.

Skin fragility and hyper-elasticity, joint laxity, easy bruising (present in our patient on more detailed history) and poor wound healing are characteristic of most subtypes. Subcutaneous spheroids (lipoma-like lesions) arise secondary to poor membrane integrity. EDS Type IV (deficient collagen III) is particularly worrisome because of tendency of spontaneous rupture of large arteries and hollow organs, such as colon and uterus. The life expectancy is significantly reduced, and women with this subtype are advised to avoid pregnancy.

In nearly 50% of patients with clinical EDS, it is hard to clearly establish a specific category. The skin biopsy is often non-diagnostic, as it was in this case. Special collagen testing by protein electrophoresis is sometimes done; in our case it was only able to exclude the vascular type of EDS (collagen III was normal).

No specific therapy for EDS exists at present. Physical exercise to strengthen muscles and provide support for the loose ligaments is beneficial. Surgical care is often complicated and requires special techniques because of poor wound healing and vascular fragility.

Case 4
Diagnosis: Dermatitis Herpetiformis

Dermatitis herpetiformis (DH) is a chronic, intensely burning, pruritic, vesicular skin disease associated in most instances with subclinical gluten-sensitive enteropathy (celiac disease) and IgA deposits in the upper dermis. The skin biopsy (H/E and IF) is diagnostic, as it was in our case. There is a strong association with specific human histocompatibility leukocyte antigens: HLA-B8 (60%), HLA class II antigens HLA-DR3 (95%), and HLA-DQw2 (100%).

Gluten-sensitive enteropathy occurs in nearly all patients with DH, and is demonstrated by small bowel mucosal biopsy. Patients with DH appear to be antigenically stimulated by dietary gluten, which results in formation of IgA-containing immune complexes leading to gastrointestinal and skin changes. Atrophy of small intestinal villi occurs and is similar to, but less severe, than that found in ordinary celiac sprue. Symptoms of malabsorption are rarely encountered (<20%). Small bowel lymphoma, achlorhydria, atrophic gastritis, and pernicious anemia are the gastrointestinal complications linked with DH.

A strong association has also been reported between DH and diverse thyroid abnormalities, including Graves’ disease, Hashimoto’s thyroiditis, and idiopathic hypothyroidism. Other autoimmune diseases, including rheumatoid arthritis, SLE, ulcerative colitis, and Sjogren’s disease are also found with DH, but are more rare.

DH usually begins in the second to fifth decade of life, presenting with intensely burning papules, vesicles, and bullae, mostly in grouped arrangement, resembling herpes simplex or zoster (thus the term herpetiformis). The vesicles are symmetrically distributed and classically appear on extensor surfaces, including elbows, knees, back, neck area, shoulders and buttocks. Dietary iodides (found in seafood) and wheat overload are classically known to exacerbate DH, as was the case in this patient.

Dapsone and sulfapyridine are the medications of choice for the treatment of DH. Patients with a classic history and vesicular eruption may be given a trial of sulfone therapy, and the dramatic relief of symptoms within hours or few days supports the diagnosis of DH. Although celiac disease in patients with DH is often subclinical, the skin lesions respond greatly to gluten withdrawal. Gluten-free diet may completely suppress the disease, and current evidence indicates that this diet restriction might need to be continued indefinitely.

Case 5
Diagnosis: Panic Disorder

A panic attack, according to the DSM-IV, involves a sudden onset of intense fear peaking in 10 minutes and associated with at least four of the following features:

1. Heart palpitations or tachycardia
2. Sweating
3. Trembling
4. Shortness of breath
5. Chest pain
6. GI upset
7. Dizziness
8. Derealization or depersonalization
9. Fear of losing control, going crazy or dying
10. Numbness or paralysis
11. Chills or hot flushes

Panic disorder involves repeated unexpected panic attacks followed by I) at least a month of persistent fear of subsequent attacks or worry about implications or consequences of the attack (i.e. fear of “going crazy” or having a heart attack) or II) a significant change in behavior (i.e. agoraphobia).

Panic disorder is a potentially debilitating condition with a lifetime prevalence of 3.5%. Many patients with Panic Disorder visit the ED repeatedly before seeking psychiatric treatment, as the disorder is difficult to recognize. According to the 1995 Montreal Heart Panic Project (MHPP), 25% of patients presenting to the ED with chest pain met DSM-IIIIR criteria for Panic disorder, though panic disorder is mentioned in less than 1% of cases as a differential diagnosis. The MHPP reported that 57% of panic disorder patients had comorbid psychiatric disorders (generalized anxiety disorder, agoraphobia, and major depression), and 25% had suicidal thoughts in the week prior to their ED visit. Once identified, panic disorder can be treated with a combination of medication (SSRIs and benzodiazepines) and cognitive behavior therapy.