Case 1: 3 year old with recurrent infections

Case History

A family doctor has referred a 3 year old boy with recurrent infections to your general pediatric practice. Mom states that he "seems to always have a cold", they last from 5-10 days and he recovers without intervention. He has had 5 ear infections in the last year, all treated with antibiotics. He has been treated with antibiotics once for "bronchitis" from a walk in clinic. He presented to the hospital ED once 2 months ago with a high fever, rhinorrhea and a cough, but was sent home without further treatment. Mom is very concerned.
RoS: Negative
PMH: Healthy, ventolin prn, no hospitalizations/sx, no allergies, IUTD
FHx: Brother with asthma, mom and dad are healthy, no consanguinity, no infant deaths
SHx: Attends day care, dad smokes in the house

On examination you find that he is 50%ile for height and weight. Vital signs are stable.
HEENT- rhinorrhea, cervical LAD, red oropharynx
Neuro, CVS, Resp, abdo, MSK – All normal

Questions

1. Could this be an immunodeficiency? What are the supporting features?

2. How many infections are patients "allowed" before you would be concerned that he may have a primary immunodeficiency?
    a. AOM
    b. Sinus infections?
    c. Pneumonia??
    d. Deep-seated infections (osteomyelitis, cellulitis, meningitis)?
    e. Abscesses?

3. What are some other red flags for immunodeficiency?

Case 2: 3 year old with recurrent infections

Case History

A family doctor has referred a 3 year old boy with recurrent infections to your general pediatric practice. He was well until 8 months of age, when he developed RLL pneumonia which resolved with a course of oral antibiotics. At 2 years of age, he was hospitalized for LUL pneumonia, and then again at 3 years of age for LLL pneumonia which required iv antibiotics. His mom states that "a bug grew in his blood" but she cannot recall what organism it was. He has had multiple ear infections starting at the age of 6 months, his mom cannot remember how many. Most recently he was treated with antibiotics for erysipelas.
RoS: Currently has a cough, a "picky eater", has always had difficulty gaining weight
PMHx: Unremarkable pregnancy, IUTD, no allergies, normal development
FHx: 2 older sisters are healthy, mom has asthma and allergies, maternal uncle died of pneumonia, maternal grandmother with pernicious anemia, dad healthy, no consanguinity, no infant deaths

Questions:

1. Could this be an immunodefiency? If so, what type? Name 3 red flags for immunodeficiency in this case?

2. What are some non-infectious signs and symptoms associated with immunodeficiencies that you should ask about in your history?

3. What are some key physical findings you will look for?

4. What investigations will you do given the information you have received so far

5. Your preceptor will give you some results. What is your diagnosis?

Case 2 continued

You have referred this patient to immunology, and together you have made the diagnosis of X-linked Agammaglobulinemia (Bruton's Agammaglobulinemia). The patient and family have come back to your office in follow up, and have several questions for you. The immunologist is on holidays, so you are on your own!

1. How did he inherit this? Does this mean all of his children will be affected?

2. Is there a cure?

3. What can be done to treat this/prevent further infections?

4. Can/should he be vaccinated?

5. What is currently the most cause of death in people with this condition?

Case 3: A 9 month old with FTT

History

The parents of a recently adopted infant have come to your pediatric clinic for their first visit. The infant is 9 months of age, and arrived to Canada from Rwanda 2 weeks ago. They are concerned that since coming to Canada, she has been "fighting of a cold", with frequent cough, and has had several subjective fevers. The parents state that they have no information about their child prior to adoption. They do know that she was recently treated for pneumonia. Developmentally she is just starting to sit on her own, she makes some noises but does not yet babble, she grasps at toys and can bring them to midline. Family history is also unknown.
RoS: Fussy child, occasional cough, eats well, no diarrhea/vomiting, good U/O

When you examine her, you find an infant who is <3rd%ile for weight, 3%ile for length. General – Tired looking, wasted appearance, nontoxic, HEENT – MMM, palpable lymph nodes, normal fontonale, CVS – Normal, Resp – Decreased AE to RLL, diffuse crackles, Abdo – Distended, MSK – Diffuse rash, dry skin.

You do some initial bloodwork. CBC shows Hb 100, WBC 11 with normal diff, platelets 175. BUN, creatinine normal. Liver enzymes normal, INR 1.6, PTT 30. Ig GAM all slightly low.

Questions:

1. What could explain this clinical picture? Are you suspicious of an immunodeficiency? If so, what is the most likely cause?

2. What are some causes of secondary immunodeficiency?

3. What further investigations would you order in this scenario, if any?

Case 4: 8 months old boy with an abscess

History

You are seeing an 8 month old boy with a perianal abscess in the ED. When taking a history, you hear that he has had an episode of lymphadenitis in the past. RoS: Decreased intake in the last 24 hrs, decreased u/o, no vomiting/diarrhea
PMH: 1 previous hospitalization (lymphadenitis), no previous surgeries, no allergies. He is not immunized. He has a history of poor wound healing. Followed closely by his family physician for FTT. Development normal
FHx: No siblings, parents are healthy. No consanguinity, no infant deaths
SHx: Dad recently lost his job, mom works at Superstore. Baby attends daycare

On examination you find an unwell looking child, fussy, inconsolable. He is 3%ile weight, 3%ile for height, fever of 38.7, HR 160, RR 30, BP 80/58. HEENT – cervical LN palpable, mucous membranes dry. CVS/Resp normal. Abdo – No palpable masses, slightly distended. MSK – Red, fluctuant mass in perianal area. Initial bloodwork shows Hb 120, WBC 18 (neuts 15, lymphs 2), platelets 280. ESR 35, CRP 120. Lytes, renal function normal.

Questions

1. What is your initial management plan?

2. Do you suspect an immunodeficiency? List the possible immunodeficiencies this could be, and why?

3. What investigations would you do to confirm your diagnosis?

4. The nurse has just called to tell you that the baby has not had any urine output in the last 24 hrs. He is on maintenance iv fluids and is feeding on top of that. After reassessing your fluid balances and clinically assessing the child, you determine that he is euvolemic. What is happening?

Case 5: Bloody diarrhea with a twist

History

You have just admitted a 10 month boy for several episodes of bloody diarrhea. In the last 2 days, mom states that he has had 3 bouts of fresh red blood per rectum. He has no fever, has not travelled has not eaten any new foods. He is well with the diarrhea. He is still eating and drinking well, and he has not had any change in his urine output. He has had no sick contacts
RoS: No resp sx, no vomiting, currently on Amoxicillin for an ear infection
PMH: History of eczema, treated with 0.1% betaderm. 6 bouts of AOM, treated with antibiotics, hospitalized once when he was 2 months of age for bronchiolitis.
FHx: Parents healthy, one healthy sister, no consanguinity, no neonatal deaths

On exam, you find a well looking child, vitals are stable. Growth is 10%ile for weight and height. HEENT – Red L TM, scarred R TM, MMM, no LAD, CVS/resp/abdo normal. MSK – eczema in flexural surfaces, petechiae to face, upper and lower extremities.

Questions

1. What is currently on your differential diagnosis?

2. What initial investigations would you order?

3. Your preceptor will now give you some results. What is the most likely diagnosis?

4. Wiscott Aldrich Syndrome is a condition affecting reorganization of the cell actin cytoskeleton. This is required for T cell migration, and T cell interaction with other target cells. Knowing this:

    a. What kind of immune dysfunction do you expect in WAS?

    b. What immune work up would you order? What do you expect to see if you did a full immune work up?

5. You are the general pediatrician looking after this patient in the community. What types of complications do you need to watch for? How would you advise the family?