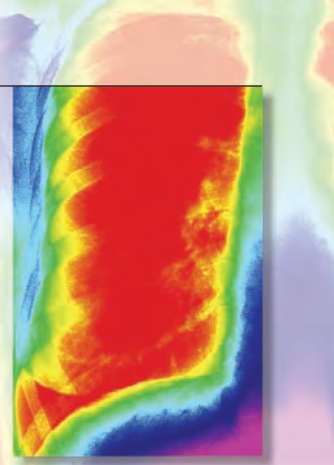


Case Study #33



Cystic fibrosis

Emily started off the first week or so of her life in what appeared to be fairly good health however, by four months of age Emily was clearly not at all well. She was underweight and she was suffering for the second time with pneumonia and was distressed. On admission to hospital, she was treated with antibiotics and her blood analysis showed the following abnormal results: hemoglobin, hematocrit, serum albumen, and total protein were all low. A sputum sample, when cultured, showed both *Corynebacterium* organisms and *Streptococcus pneumoniae*. It was also noted that her stools were bulky and greasy looking.

antibiotics and new sources of infection are another complication.

A critical role for Emily's caregivers is to help maintain drainage from the bronchi. This involves placing her at the edge of the bed with her head down and using a device used to "percuss" her chest so as to allow the thick secretion to drain. This treatment may take half an hour, twice a day to be effective. In addition, there are several medications that may be prescribed to help maintain respiratory function.

The nutritional deficiencies that are caused by blockage of the pancreatic duct are managed by giving the person vitamin supplements and capsules containing diges-

become thick. The organs that are most seriously affected are the lungs and the digestive system (especially the pancreas). In the case of the lung secretions, the chloride concentration is increased and, with the pancreas, the various digestive enzymes that it would normally pass into the digestive system are blocked.

Cystic fibrosis is inherited through an autosomal recessive gene mutation. (Several different mutations of this gene have been discovered.) Autosomal is the term which is used to indicate the chromosome involved is neither X nor Y – the sex chromosomes. In cystic fibrosis the gene mutation is located in chromosome 7. Recessive inheritance means that, for the disease to be expressed, the mutation must be acquired from both the mother and father. In contrast, a dominant mode of inheritance would require only one of the two genes to be mutated. In the case of diseases caused by recessive modes of inheritance, should the child inherit one gene that is mutated, but another that is normal, then the child will be described as being a carrier. In the case of cystic fibrosis, a carrier would not have the disease. (As noted earlier, there are several different mutations that can occur, and depending on which one is involved, so may vary the severity of the disease.)

Starting in 2008, a screening test for cystic fibrosis will be added to the battery of tests used to screen all newborn children in Ontario. Early diagnosis of the disease and appropriate early treatment is expected to reduce the complications that arise from the disease. See the internet for more information from the Cystic Fibrosis Society (<http://www.cysticfibrosis.ca>). ❖

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A provisional diagnosis of cystic fibrosis was made. (Her sister, who was three years older, has been in good health since birth.) The diagnosis was confirmed by doing a sweat chloride test. Her first result was 72 mmol/L and a second test was 90 mmol/L. The upper limit of normal is 50 mmol/L.

Subsequently, Emily's treatment and management was aimed at dealing with the excessively thick secretions that her lungs were producing, and in dealing promptly with any lung infections. Repeated bouts of lung infection may result from the bacteria becoming resistant to previously effective

enzymes. There is no cure for the disease and the intensive therapy that is required to prolong the patient's life will likely take a heavy toll on both the patients and their caregivers. However, as more is learned about the disease and its complications, the patient may be expected to live into his/her thirties.

Discussion

Cystic fibrosis is an inherited condition where there is a mutation of the gene that regulates the passage of sodium chloride through the body's cell walls. As a result, fluids that would normally be thin enough to act as a lubricant

Answers to page 22 Crossword Puzzle

<p>28. Blood 29. Accuracy 32. Temperature 34. Results 37. Confrontation</p>	<p>13. CMLTO 16. Graph 17. Delay 20. Check 22. Knowledge 25. Airborne 26. Fax 27. SARS</p>	<p>Down 1. Universal 3. Lunch 5. Bird 7. Unique 8. Needle 9. Wait 12. Washing Hands</p>	<p>30. Mask 31. STAT 33. Wrist-band 35. Screening 36. Specimens 38. Gloves 39. Communication</p>	<p>14. Sleeve 15. Flagged 18. Quality Control 19. Finger-print 21. Dipstick 23. MRSA 24. Explain 28. Bio-Hazard</p>	<p>2. On call 4. Helpline 6. Humour 8. Normal 10. Label 11. Mix 13. Chart</p>
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