CASE THREE

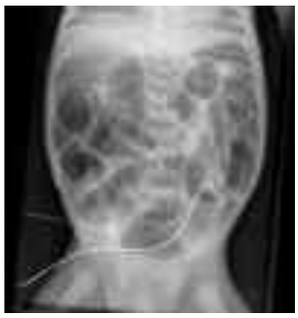
Short case number: 3_6_03
Category: Children & Young People
Discipline: Paediatrics Medicine
Setting: Rural_Hospital
Topic: Cystic Fibrosis_Meconium Ileus

Case

You are the intern covering the neonatal nursery, Georgie Taylor is 12 hours old. The nursing staff have asked you to review her because she has not passed meconium. She has not been feeding very well and has just vomited. On assessment you note that Georgie has a distended abdomen.

Questions

1. You are concerned that Georgie may have a bowel obstruction, and you organise for an abdominal x-ray to be done [films shown below]. What are the features seen on the abdominal radiograph?



2. The radiologist reports that the findings are consistent with meconium ileus. You speak with surgeon who explains that Amelia, will need to be transferred to the 'children's hospital' and that he will come and speak with parents. The surgeon explains the condition and the likelihood that Amelia has cystic fibrosis. Her father asks what this is and how did Amelia 'get it'? What would you explain to the parents about Cystic Fibrosis?
3. Explain the current newborn screening testing that is performed for cystic fibrosis and outline the limitations of this test.
4. As you are preparing Amelia for transfer to the children's hospital you think about what lies ahead for her and her family. Summarise in the form of a management plan the main problems that can occur in Cystic fibrosis and outline the key aspects of management.

Resources

- South M, Isaacs D editors. Practical Paediatrics. 7th edition. Edinburgh: Churchill Livingstone; 2012.

ANSWERS

Question 1

You are concerned that Georgie may have a bowel obstruction, and you organise for an abdominal x-ray to be done [films shown below]. What are the features seen on the abdominal radiograph?

X-ray shows:

- Distended loops of intestine with thickened bowel wall. (As seen on this x-ray)

Other X-ray signs include:

- A large amount of meconium mixed with swallowed air producing the so-called 'ground-glass' sign typical of meconium ileus. This is a characteristic feature but often absent.
- Calcification, free air or very large air-fluid levels suggesting bowel perforation.

Question 2

The radiologist reports that the findings are consistent with meconium ileus. You speak with surgeon who explains that Amelia, will need to be transferred to the 'children's hospital' and that he will come and speak with parents. The surgeon explains the condition and the likelihood that Amelia has cystic fibrosis. Her father asks what this is and how did Amelia 'get it'? What would you explain to the parents about Cystic Fibrosis?

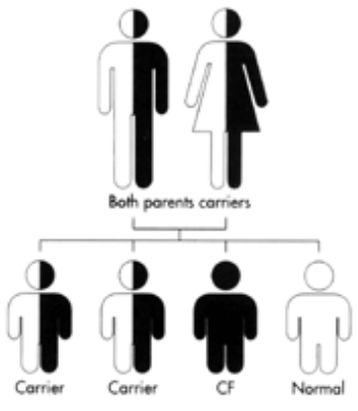
Cystic fibrosis is the most common life-threatening autosomal recessive disorder in Australians, affecting approximately 1 in every 2500 births. It is caused by a defect in the cystic fibrosis transmembrane conductance regulator gene (*CFTR*). The *CFTR* gene encodes a protein for a cyclic adenosine monophosphate (cAMP)-regulated chloride channel present on many epithelial cells, including those of the conducting airways, gut and genital tract. The commonest mutation, Δ508, accounts for approximately 70% of mutant alleles and more than 1300 mutations have been described.

Cystic Fibrosis (CF) is an inherited recessive genetic condition. Amongst people of Caucasian ancestry 1 in 25 are genetic carriers for CF, usually without knowing it.

If a baby is born with CF, it means that both parents are genetic carriers for CF. However, even if they are genetic carriers, there is no guarantee that the child will be born with CF.

If two people are genetic carriers for CF and they have a child there is (with every pregnancy):

- A 1 out of 4 (25%) chance that the child will have CF
- A 2 out of 4 (50%) chance that the child will be a genetic carrier for CF
- A 1 out of 4 (25%) chance that the child will not have CF and will not be a genetic carrier for CF.



Question 3
Explain the current newborn screening testing that is performed for cystic fibrosis and outline the limitations of this test.

In Australia all babies are screened at birth for Cystic Fibrosis. A small blood sample is taken about three days after birth and if this test proves to be positive genetic testing of the CFTR gene will be done. Finally a sweat test will be carried out to measure the amount of salt in the sweat and it is with this test that a diagnosis will be made.

While newborn testing picks up almost all babies with CF (about 95%) the test will miss some. A sweat test should be arranged if there are phenotypic features suggestive of cystic fibrosis

Sweat Test:
The diagnosis confirmed with a sweat test (pilocarpine iontophoresis) at 6-10 weeks. An elevated sweat chloride and sweat sodium is diagnostic.

Question 4
As you are preparing Amelia for transfer to the children's hospital you think about what lies ahead for her and her family. Summarise in the form of a management plan the main problems that can occur in Cystic fibrosis and outline the key aspects of management.

The goal of treatment is to maintain as high a quality of life as possible for as long as possible in order to slow the relentless progression of lung disease that occurs in cystic fibrosis.

Respiratory management:

- Prompt use of antibiotics to delay the onset of bacterial colonization.
- Aggressive treatment of recurrent respiratory infections.
- Promotion of mucociliary clearance by daily physiotherapy.
- Minimization of other causes of lung damage (e.g. smoking, aspiration).
- Promotion of normal growth through high-energy diet and pancreatic supplementation.
- Identification and treatment of complications as they arise (asthma like disease, allergic bronchopulmonary aspergillosis (ABPA), haemoptysis, pneumothorax, etc.).

Gastroenterological and nutritional management:

- Pancreatic enzyme replacement (lipase, protease, amylase) at each meal.
- High energy diet.
- Vitamin supplementation with vitamin A, D, E and K, and salt tablets.

Psychological management:
Cystic fibrosis is a lifelong chronic condition. As children grow and mature into adolescents and young adults, the psychosocial aspects of the disease take on different dimensions for individuals, siblings and parents. In adolescence, attention to body image issues and feelings of difference due to chronic disease can help maintain young people's adherence with the health-care regimen. Declining health despite good adherence can be especially demoralizing.