# Case 4 The blue baby

*Mrs Smith has brought her baby to A&E because she says 'he has started turning blue'.*

## What are your immediate differential diagnoses?

1 *Respiratory causes*:
- Congenital respiratory disorder.
- Acquired respiratory problem.
- Congenital obstruction.
- Acquired obstruction.

2 *Congenital cyanotic heart disease*:
- Tetralogy of Fallot.
- Transposition of the great arteries.
- Other rarer cyanotic defects.

3 *Lack of respiratory drive*:
- Seizure disorder.
- Congenital CNS malformation.
- CNS infection.
- Drugs.

## What would you like to elicit from the history?

### Demographics

- Exactly how old is this baby? Congenital problems are most likely to present in the first month, cyanotic heart disease when the duct closes after 3–7 days.

### Presenting complaint

Find out more about the blueness:
- Where has the mother noticed the colour change (peripheral or central)?
- How suddenly did it come on?
- What has the mother noted about her child's breathing – is it laboured, erratic or normal?
- What was the baby doing at the time she noticed the colour change?
- Are there any associated symptoms (i.e. crying, difficulty breathing, general distress, unusual quietness or movements)?

### Systems enquiry and past medical history

- Does the child have any medical conditions?
- Has the child had any breathing problems before?
- Do the parents have any concerns about the child's development?
- Are there any other signs of infection – poor feeding, fever, sleepiness?

### Family history

- Are there other family members with congenital heart problems?
- Are the parents related?

### Obstetric history

- Were there any problems at birth, e.g. meconium-stained liquor?
- Was the child born at term?
- Were there any problems with antenatal scans?

*Baby Smith is only 3 days old. He was born at home, at term, and everything had seemed fine until he started turning blue around the mouth a couple of hours ago. Mrs Smith put this down to him crying at the time but it did not seem to improve when he calmed down.*

*He was feeding well at the breast, but has not seemed interested today. He is breathing a little faster than normal, but does not seem to be struggling.*

*Mrs Smith was well throughout the pregnancy and neither she, nor her husband, has any relevant past medical history. All the scans were normal in pregnancy.*

## Review your differentials

1 *Respiratory causes*:
- *Congenital respiratory disorder*: It can be difficult to tell respiratory causes from cardiac causes of cyanosis in one this young. Investigations are required. The normal work of breathing is against a respiratory cause, but will not exclude it.

- *Acquired respiratory disorder*: Baby Smith may have a chest infection, or bronchiolitis, but the lack of respiratory symptoms makes this unlikely.
- *Congenital obstruction*: Upper airway obstruction is unlikely without stridor (see Case 13, p. 67, for further discussion of upper airway obstruction).
- *Acquired obstruction*: This might be caused by a foreign body. However, baby Smith is too young to put things in his mouth. This is, therefore, an unlikely diagnosis, but it is not out of the question (see Case 23, p. 106). He could also have an airway infection, such as croup, but again this is unlikely at this age.

2 *Congenital cyanotic heart disease*:
- *Tetralogy of Fallot*: This can present soon after birth in extremely severe cases. However, the majority of children present with hypercyanotic 'spells' during the first year of life. These episodes, usually accompanied by crying, distress and apparent air hunger, are characteristic.
- *Transposition of the great arteries* (TGA): In this disorder, the pulmonary artery is connected to the left ventricle and the aorta to the right ventricle. This gives rise to two separate parallel circulatory systems. In fetal life and the first few days *ex utero* the natural shunts created by the foramen ovale and the ductus arteriosus allow mixing of the two circuits. When the ductus closes, the infant cannot get oxygenated blood to the body.

## What will you do now?

This is an emergency. Cyanosis indicates a potentially life-threatening lack of oxygenated blood. You must assess the baby and decide what degree of resuscitation is required. Check the airway for obstruction, and assess breathing and circulation. Check pulse rate and perfusion.

Further examination must be rapid – what key things will you look for?

- *General observations*:
  - Cyanosis – central or peripheral?
  - Level of arousal – is this baby's conscious level decreased? If it is, it implies a critical level of hypoxia.
- *Close assessment of the respiratory system*:
  - Assess work of breathing.
  - Percuss and auscultate the lung fields.
- *Close assessment of the cardiovascular system*:
  - Check capillary return.
  - Auscultation for murmurs.
  - Feel the femoral arteries.
  - Check BP.

> *On examination, there is deep peripheral and central cyanosis. The baby is tachypnoeic with minimally increased work of breathing. There are no apparent obstructions in the mouth or pharynx.*
>
> *The HR is 165, capillary refill time 2–3 s, BP 65/35 and the femorals are palpable. The chest is moving well and clear to ausculation and percussion.*

## What investigations will you perform?

- Arterial blood gas.
- Chest X-ray.
- Nitrogen washout/hyperoxic test.
- ECG.
- Echocardiography.

> *Some of the results are back:*
>
> $PaO_2$ *3.2 kPa*
> $PaCO_2$ *4.5 kPa*
> *pH 7.15*
> *BE –7.1*
> *The chest radiograph is also back. The ECG is normal.*

## What do you make of the blood results and chest film?

There is a metabolic acidosis. This is due to lactic acidosis resulting from poor tissue oxygenation, despite reasonable cardiac output. The chest film (Fig. 8), demonstrates the classic 'egg on its side' cardiac shadow.

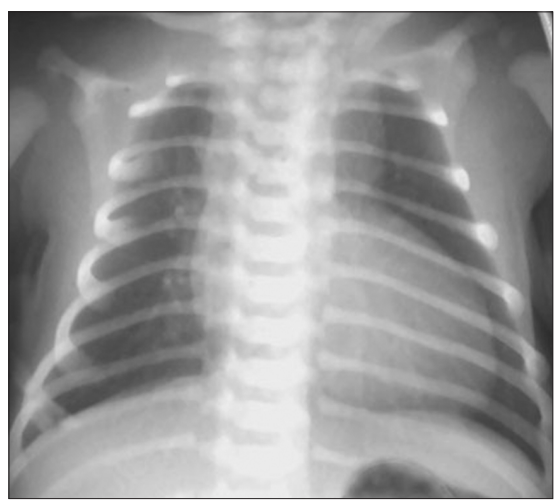

**Figure 8** Baby Smith's chest radiograph.

## What is the nitrogen washout test and what do you expect it to show?

This test differentiates cardiac from respiratory causes of cyanosis. The child is placed in 100% $O_2$ for 10 min. This maximally oxygenates the pulmonary circulation. Therefore, if the origin of the problem is respiratory, the desaturation should be ameliorated (unless extremely severe) as the extra alveolar oxygen overcomes the diffusion problem in the lungs. If the child is still cyanotic by the end of the test there must be a right-to-left cardiac shunt, as the increased oxygen is unable to get into the systemic blood because of abnormal plumbing.

> *Baby Smith's 10 min blood gas showed a $PaO_2$ of 7 kPa (an arterial $PaO_2$ of less than 20 kPa is considered to indicate a high likelihood of cyanotic heart disease).*

## What do you expect the echocardiography to show?

The echo would show the anatomical cause of the problem. However, neonatal echo is only available in specialist centres. You will have to stabilize the child before you can get an echo.

## What is the management plan?

It is imperative to maintain some link between the right and left circulations. Cyanosis is developing now because the ductus arteriosus is in the process of closing. This is a 'duct-dependent' circulation. To maintain patency of the duct you administer a continuous prostaglandin E2 infusion (in extreme cases the intraosseus route is acceptable), which relaxes the ductus smooth muscle.

You continue supportive measures, concentrating on:

- Temperature.
- Correcting the acidosis.
- Setting up a dextrose infusion to maintain blood glucose.

Call the cardiology team to arrange transfer to a paediatric cardiology centre.

## What is the definitive treatment?

First the child must have an anatomical diagnosis. The echo confirms transposition of the great arteries. The aorta and pulmonary arteries are in parallel with the aorta in front of the pulmonary artery. Fortunately for baby Smith, ultrasound also shows a large ventricular septal defect. Septal defects are often associated with TGA and make the presentation less severe.

Surgical correction must be performed.

- The Rashkind septostomy is a temporary measure to allow mixing of the two circulations by removing the atrial septum. This can be done by catheter in an ICU.
- The treatment of choice is the arterial switch operation. If successful, this has the fewest long-term complications because it is closest to the natural anatomical structure. The arteries are transected and replaced (note that the coronary arteries also have to be replaced). This must be done within the first 2 weeks of life, before pressure changes in the heart begin to detrimentally affect the myocardium (the left ventricle regresses due to attachment to the low pressure pulmonary circulation).
- Sometimes a 'Mustard' or 'Senning' repair is usually done at about 6 months of age. The aim here is to create a longer term solution by using a baffle to redirect caval blood through the mitral valve (and therefore the left ventricle and *pulmonary* artery). Pulmonary return is redirected to the tricuspid valve. This tends to result in heart failure in the long term because the right ventricle does not have the blood supply to cope with the work of supplying the high pressure systemic circulation.

## What is the prognosis?

The arterial switch operation has a mortality of about 5% but success rates are improving all the time. If it is successful, the prognosis is good. Since these techniques were developed fairly recently, it is difficult to say what the very long-term complications may be. There is a risk of residual pulmonary stenosis, which may lead to right heart failure.

## Comments

- The body is not as concerned about hypoxia as it is about acidosis or hypercapnia, causing tachypnoea and increasing tidal volume. Disease in the lungs also directly stimulates the respiratory centre. The effect of this is that cyanosis caused by a cardiac malformation produces few respiratory signs, but that from a respiratory cause produces florid signs.
- *VACTERL*: The traditional VATER acronym (vertebral anomalies, anal malformations, tracheo-oesophageal fistula and radial and renal abnormalities) is sometimes extended to include cardiac and limb deformity. This is a well recognized group of anomalies. Three or more must be present in order to apply this term.
- *Tetralogy of Fallot*, which includes:
  - An overriding aorta.
  - Right ventricular outflow tract obstruction.

- Ventricular septal defect.
- Right ventricular hypertrophy.

• This is often associated with syndromes (such as Noonan's, Down's, maternal phenylketonuria). The tetrad results in mixing of oxygenated and deoxygenated blood, a reduction in pressure in the pulmonary artery and elevated pressure in the right ventricle (Figs 9 and 10). The older child with an uncorrected defect may be seen to adopt a characteristic squatting position. This obstructs left ventricular output, increasing LV pressure and forcing more blood through the obstructed RV outflow tract.

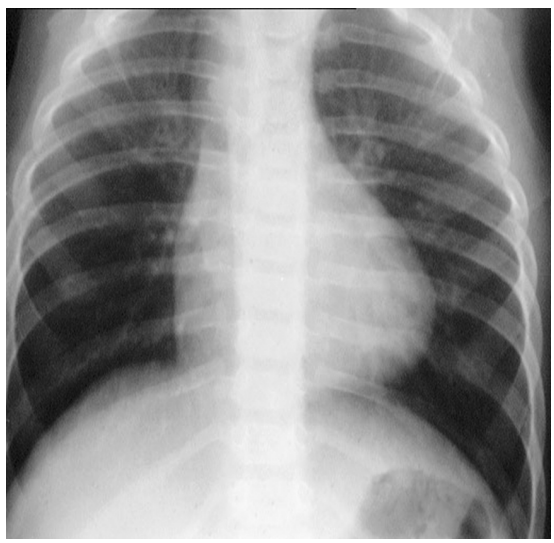

**Figure 9** Radiograph from a child with Terralogy of Fallot. Note prominent right ventricle and pulmonary oligaemia.

• An ECG at birth shows right axis deviation and right ventricular hypertrophy in all babies. Normally this changes over the first 5 years to an adult pattern. With RV hypertrophy this does not happen. The ECG is not usually helpful in the diagnosis of anatomical heart disease unless conduction is affected.

• In Fallot's, the CXR may show a characteristic 'boot-shaped' heart. Definitive diagnosis is on echocardiography.

• Without treatment most die before their teens. With surgery, 95% survive into the third decade. Surgical repair is in two stages. An early palliative procedure is the Blalock–Taussig shunt (look for the scar behind the left or right scapula). This joins the subclavian artery to the pulmonary artery. When the child is older, final reconstructive surgery can be undertaken.

• *Incidence rates*: Overall, eight in 1000 live born babies will have a heart defect. Most of these are asymptomatic. Many of the septal defects close in their own time.

- Ventricular septal defect (Fig. 11) 33%
- Pulmonary stenosis 8%
- Persistent ductus arteriosus 12%
- Atrial septal defect 6%
- Coarctation of the aorta 6%
- Aortic stenosis 5%
- Tetralogy of Fallot 6%
- Transposition of great arteries 5%

## Further reading and information

British Heart Foundation, http://www.heartstats.org. The British Heart Foundation has useful information on the incidences of congenial heart disease.

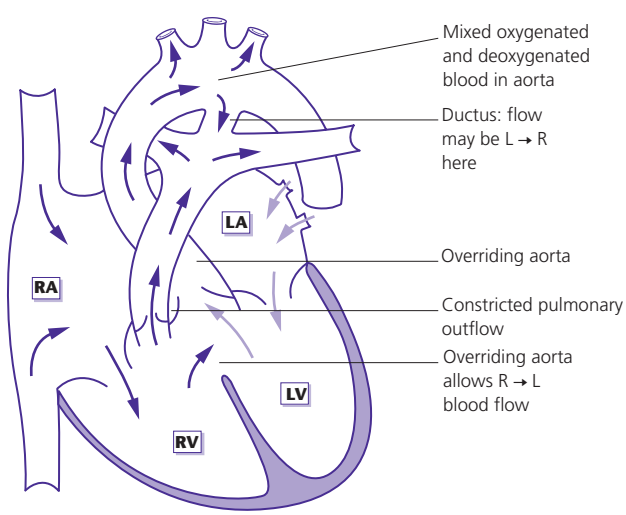


**Figure 10** Tetralogy of Fallot.

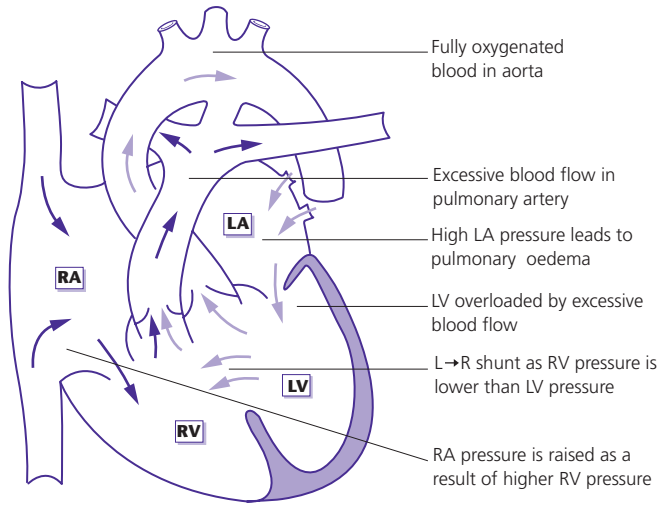


**Figure 11** Ventricular septal defect. LA, left atrium; LV, left ventricle; RA, right atrium; RV, right ventricle.

Tanner K. Cardiovascular malformations among preterm infants. *Paediatrics* 2005; **116** (6): 833–8.

Treasure T. Congenital heart disease. *British Medical Journal* 2004; **328**: 594–5.

## CASE REVIEW

Baby Smith is 3 days old when his mother notices that he seems to be becoming increasingly cyanosed, although he has little respiratory difficulty. A cardiac diagnosis rather than a respiratory one is suggested by a minimal increment in $PaO_2$ when put into 100% oxygen. His chest radiograph shows no respiratory disease and he has an abnormal cardiac silhouette. He is sent to a paediatric cardiac centre for a diagnostic echocardiogram and corrective surgery. Despite the difficulty and risks of the procedures he will undergo, his outcome is likely to be good.

## KEY POINTS

- Cyanosis is cardiac or respiratory in origin.
- The child's age, and the presence or absence of associated symptoms, are useful in directing the diagnostic process.
- Cardiac abnormalities are relatively common (eight in 1000 live births).
- Prostaglandins are used in the immediate management of patients with duct-dependant circulation (to maintain patency of the ductus arteriosus).
- Surgical correction is the definitive treatment for both transposition of the great arteries and tetralogy of Fallot.