

48. THALASSAEMIA AND SICKLE CELL ANAEMIA

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48.2 Thalassaemia

48.2.1 Clinical Description

The thalassaemias are hereditary anaemias due to abnormalities in the synthesis of haemoglobin (the oxygen-carrying protein in red blood cells). The severe effects of thalassaemia, which because of treatment are rare in the UK, are the result of severe anaemia which stimulates the production of red blood cells; this expands the bone marrow mass resulting in osteoporosis (thinning of the bone) with the potential for spontaneous fractures. Retardation of bone growth, marked enlargement of the liver and spleen and a tendency to acute crises occur. For example infection can cause a catastrophic fall in the number of red blood cells. Modern treatment makes these features unusual.

48.2.2 Care Needs

- (i) Correction of the anaemia by regular blood transfusion, usually every 6 to 8 weeks, avoids the complications and allows the child to develop normally. As a result of regular blood transfusions iron overload becomes a problem because humans have a limited ability to excrete iron. This can be overcome by use of the iron chelating drug, desferrioxamine, which is administered under the skin by an infusion pump. The infusion is given over a period of several hours during the night. The pump is set up before the child goes to bed and the needle removed first thing in the morning. Each of these actions normally takes no more than 15 minutes.
- (ii) Although the pumps are reliable and the occasional failure would not have a significant effect on the course of the disease, it would not be unreasonable for a parent to look in on the child several times through the night whilst this therapy is in progress. Consequently, unless it is evident that the child has completely adapted to the presence of the pump, watching over by night may be considered appropriate. Children have usually adapted to the presence of the pump by the age of 12. Beyond this age, they would normally not be expected to have care or mobility needs in excess of those of a healthy child unless they had developed any of the complications of the disease. The care needs would then depend on the nature of the complications and the severity

of the symptoms produced.

48.2.3 Mobility Considerations

Thalassaemia does not in itself cause any problems with walking.

48.3 Sickle Cell Anaemia

48.3.1 Clinical Description

- (i) Sickle cell anaemia is an inherited disorder of haemoglobin (the oxygen-carrying protein in red blood cells). To develop sickle cell anaemia, a person must inherit a gene for the abnormal protein from each parent. People with only one such gene are said to have sickle cell trait. Only in exceptional circumstances would a person with sickle cell trait develop problems.
- (ii) In sickle cell anaemia, the abnormal haemoglobin is less soluble than the normal form and this causes deformity (sickling) of the red blood cells in certain circumstances. The effect is to plug the arterioles and capillaries, depriving tissues of their vital oxygen supply. As a result tissue damage occurs.
- (iii) These episodes are called "crises". During a crisis, a person with sickle cell anaemia may experience fever, abdominal pain, vomiting and severe pain in the long bones, back and joints. The person often becomes suddenly and severely anaemic. Other features may include stroke, paralysis of the cranial nerves (nerves controlling the head muscles), chronic ankle ulcers and avascular necrosis (death of bone) at the top of the thigh. Repeated crises lead to a chronic deterioration in general health. In adults there is a severe (though variable) anaemia, and progressive lung and kidney failure. As a result, life expectancy may be reduced.

48.3.2 Care Needs and Mobility Considerations

- (i) In the early years of the disease, during and immediately after an acute sickle cell crisis, the person is very ill, unable to walk and requires high levels of care. However, the individual crisis is self-limiting, and will remit within a week or so. Between crises the person will be expected to function normally, so it is important to gauge the severity, frequency and duration of crises.
- (ii) As the disease progresses, while the acute crises may continue on a regular basis, the toll on general health becomes evident. This affects both mobility and care needs. For example, avascular necrosis at the top of both thighs may seriously impede the ability to walk, whilst chronic pain, fever and general debility will generate increased care needs.

48.3.3 Duration of Needs

Whilst it is true to say that sickle cell disease is generally progressive, the condition may remain static over several years.

48.3.4 Further Evidence

In determining the severity, frequency and duration of sickle crises, a factual report from a GP or hospital medical or nursing specialist attendant may be helpful. In chronic disease where general health has become impaired, a visit from an examining medical practitioner may be required in order to assess fully the needs of the person with the disease.