

43. METABOLIC DISORDERS IN CHILDREN

43.1	Contents:	Paragraph
	Introduction	43.2
	General	43.3
	Care Needs	43.4
	Mobility Considerations	43.5
	Duration of Need	43.6
	Further Evidence	43.7

43.2 Introduction

43.2.1 Metabolic disorders (inborn errors of metabolism) are due to inherited single enzyme defects. The loss of normal enzyme activity causes a block in the metabolic pathway which in turn may produce a characteristic picture of disability. About 200 different examples are known. Some of these have no disabling effects of importance, others cause varying degrees of disability, and some may be incompatible with life. In most cases, the change in the structure of the enzyme either reduces or abolishes its effects. In other cases no enzyme at all may be produced.

43.2.2 These conditions are rare but in many instances cause severe disability, particularly severe learning disability, so they are important because of the care and mobility needs that can arise. They have a variety of effects, depending on the metabolic pathway affected, for example, haemophilia (absence of a blood clotting factor), albinism (absence of skin pigment and impaired vision) and phenylketonuria (inability to metabolise the amino acid phenylalanine which can result in brain damage).

43.2.3 The cause of these conditions cannot be corrected yet so treatment is aimed at their effects. For example, haemophilia is treated by replacing the missing clotting factor; albinism is helped by avoiding the damaging effects of bright sunlight; the damaging effects of phenylketonuria are prevented by restricting the amount of phenylalanine in the diet.

43.3 General

43.3.1 This section will deal with the care and mobility needs that arise from those conditions that require dietary restrictions as these form quite a large group with similar effects and needs. Other conditions such as haemophilia are dealt with separately [Chapter 47].

43.3.2 The classic example of an inborn error of metabolism treated by dietary

restriction is phenylketonuria (PKU). Children with this cannot break down the amino acid phenylalanine. Testing of all newborn babies for this condition and dietary restriction for those found to be affected means that now the vast majority grow up without any intellectual impairment at all. For many it is possible to relax the diet during adolescence. Women of childbearing age who wish to become pregnant will need to pay particular attention to their diet. For some with PKU and for those suffering from other disorders, dietary restriction is not so successful. The most serious effects of these conditions are on the developing brain, leading to learning disability, behavioural disturbances and epileptic fits.

43.4 Care Needs

43.4.1 Those with PKU who have been successfully treated from birth will develop normally, both physically and intellectually. Those treated less successfully will have the same care needs as any other child with severe learning disability [Chapter 35], behavioural disturbances [Chapter 36] or fits [Chapter 34].

43.5 Mobility Considerations

43.5.1 Children who have been treated for the condition since birth will develop normally and have no problems with walking. Children whose untreated disease has led to learning disability may have mobility needs as already described in Chapter 35.

43.6 Duration of Need

43.6.1 The effects of these conditions are irreversible so where there are care and mobility needs these will be lifelong.

43.7 Further Evidence

43.7.1 Many children will be attending a Child Assessment Unit which would be able to provide a report on their overall development.