Disease Cards

The Royal College of Pathologists Pathology: the science behind the cure

Congenital hypothyroidism (CHT)

How many are affected?

1 in 4,000 babies born in the UK has congenital hypothyroidism (CHT).

What is it?

Babies with this inherited condition do not have enough of the hormone thyroxine, because of an underactive thyroid gland. This hormone is needed for growth, and as a result babies with CHT do not grow properly and can develop physical and mental disabilities.

How can screening help?

Screening means that babies with the condition can be treated with thyroxine tablets to prevent serious disability. It is often too late to give thyroxine and prevent disability in an unscreened baby found to have CHT later in life. This early treatment can prevent serious illness and allow babies to live longer, healthier lives.



Phenylketonuria (PKU)

How many are affected?

1 in 10,000 babies born in the UK has phenylketonuria (PKU).

What is it?

Babies with this inherited condition cannot process the amino acid, phenylalanine, in their food. If untreated, they will develop irreversible mental disabilities.

How can screening help?

Screening means that babies with the condition can be treated with a special low protein, high fruit and fibre diet. It is often too late to give a special diet and make a difference to an unscreened baby found to have PKU later in life. This early treatment can prevent serious illness and allow babies to live longer, healthier lives.





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Medium-chain acyl-CoA dehydrogenase deficiency (MCADD)

How many are affected?

1 in 10,000 babies born in the UK has MCADD.

What is it?

Babies with this inherited condition cannot break down fats to make energy for the body. They can become seriously ill, or even die.

How can screening help?

Screening means that babies with the condition can be recognised early, their diet can be modified to contain high energy foods and parents can make sure they are eating regularly. This early treatment can prevent serious illness and allow babies to live longer, healthier lives.



Sickle cell disease (SCD)

How many are affected?

1 in 1,900 babies born in the UK has sickle cell disease (SCD) or sickle cell anaemia.

What is it?

Babies with this inherited condition have sickle-shaped red blood cells, which can block small blood vessels. This causes pain, serious infections, and even death.

How can screening help?

Screening means that babies with the condition can receive immunisations and antibiotics. This early treatment can prevent serious illness and allow babies to live longer, healthier lives. illness and allow babies to live longer, healthier lives.



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Cystic fibrosis (CF)

How many are affected?

1 in 2,500 babies born in the UK has cystic fibrosis (CF).

What is it?

Babies with this inherited condition produce excess sticky mucous which affects the lungs and digestive tract. Babies with CF may not gain weight, have regular chest infections, shortness of breath and coughing.

How can screening help?

Screening means that babies with the condition can be treated with a high-energy diet, medicines and physiotherapy. This early treatment can prevent serious illness and allow babies to live longer, healthier lives.



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