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**Cockayne syndrome (CS) – genetic testing**
This leaflet explains more about genetic testing for CS including the benefits, risks and any alternatives. If you have any more questions, please speak to a doctor or nurse caring for you.
Your doctor has requested a blood test and/or a skin biopsy for genetic testing in order to find out if your child has the rare genetic condition Cockayne syndrome (CS).

**What is genetic testing for CS?**
This test is done to confirm if your child has CS, by taking samples which are then looked at to see if there is a change in the gene that causes CS.

This is a unique test, available on the NHS, and has been specially developed to support the National CS Service.

**Blood test**
- Your child will need to have a simple blood test - numbing cream or spray can be used in order to minimise pain.
- The genetics test is then performed on this blood sample.
- The test is extremely complex and can take up to 6 months to be completed.
- The test may need to be repeated on a new sample of blood if the results are unclear.

**Skin biopsy**
- Your child will need to have a simple procedure where a small piece of skin (about 2mm) is taken from their buttock. This will be done with a local anaesthetic so it should not hurt once the skin has been numbed.
- A small stitch may be needed to close the wound.
- The skin sample will then be sent to the lab where they will grow more cells. Genetic testing will then be done on these cells.
- It usually takes about 3 months to get the results of this test.
- The skin sample will be stored in case any future testing is required.

**Basic genetics and CS**
CS is a recessive genetic condition.

This means that two copies of an altered gene must be present in order for the condition to develop.

Genes come in pairs. One gene comes from the mother and the other from the father. Recessive inheritance means both the genes from each parent must be affected to cause the disorder. People with only one affected gene in their pair are called carriers and they do not have the condition. However, they can pass the affected gene on to their children.

If you are born to parents who both carry the same affected gene, you have a 1 in 4 (25%) chance of inheriting both affected genes and developing the disease. You also have a 1 in 2 (50%) chance of being a carrier for the condition, and a 1 in 4 (25%) chance of not being affected at all.

Recessive genetic conditions
Both parents with the affected CS gene

Child who has not inherited the CS gene and does not have CS

Children with only one CS gene are carriers. They do not have the condition or symptoms but can pass CS to their children, if their future partner has the same affected gene

Parents who are closely related to each other, such as first cousins, are more likely to have children with recessive conditions. This is because they are more likely to share some of their genetic information and carry the same faulty genes. This is the case for all recessive conditions, not just CS.
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