As well as these specialists, you will see your CS nurse practitioner at clinic. Their role is to support you and your family and to oversee your local care, making sure that all services are in place. They will also work with you to undertake a comprehensive nursing assessment and put in place emergency care plans. You will be offered a yearly home visit with the CS nurse as well as telephone and email support.

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Cockayne syndrome (CS) - diagnosis
This leaflet explains more about CS, how it may affect your child, and support available.
A diagnosis of CS can leave you and your extended family with many questions. When you are diagnosed there is often a lot of information to understand. This leaflet aims to give you some basic information so that when you are ready to accept and move forward with what this means for your family, you have a starting point.

What is CS?
CS is a recessive genetic condition. This means that both parents must carry the faulty gene in order for a child to be affected. It is very rare - only 2-3 people per million worldwide will be affected with CS.

Will all patients with CS be affected in the same way?
As with many other illnesses, there is a spectrum (range). Patients with CS will have very similar attributes, but the severity of the condition will vary depending on how the genes are affected.

How will CS affect your child?
CS has some physical characteristics which are often similar across many affected patients.
- Height – this will depend on the severity of their condition
- Failure to gain weight - this may make your child’s eyes appear sunken and their noses more prominent
- Microcephaly (a small head)

Your child’s hearing and sight may be affected. This can often be managed with glasses and hearing aids. In some cases patients with CS will have cataracts which can be surgically removed.

People with CS are more at risk from tooth decay, so particular care will need to be taken with oral hygiene and making sure that a healthy diet is followed.

Your child may have problems with eating and drinking. These can be managed in several different ways ranging from thickening oral fluids, to a gastrostomy (a feeding tube into the stomach). The options will depend on the advice of the paediatrician/dietitian.

Photosensitivity (sensitivity to the sun) and photophobia (sensitivity to light) can be an issue for patients with CS. High factor (50+) sunscreen should be used if they are going out in the sun but, where possible, you should try to keep your child in the shade. Make sure that sunshades are used on car windows where appropriate. Sunglasses will be necessary, maybe even indoors, if your child suffers with photophobia.

CS will have an impact on your child’s developmental abilities. You may have noticed that they are not reaching their milestones at the same rate as other children their age. This is normal for patients with CS and will become more apparent as they get older.

Your child may develop a tremor (twitching in the limbs) and/or contractures (limbs that remain in a bent position). These may be treated with physiotherapy and medication, and should not cause distress to your child. It is possible that some patients with CS will have seizures which may require medication to manage.

CS is a life-limiting condition. This means that your child will not have an average life expectancy. There is no specific life expectancy as it will depend on the severity of your child’s condition.

How will your child’s condition be managed?
In order to manage all of your child’s symptoms there will be many different professionals involved. Each professional will have a different role in making sure that your child is kept as well and comfortable as possible.

Locally your child should have a paediatrician and a community nursing team (where appropriate) alongside your GP, dentist and health visitor/school nurse.

You will have the option to be seen each year with your child at the multidisciplinary CS clinic at St Thomas’ hospital. This will give you the opportunity to see a range of specialist consultants in CS genetics, dental, dermatology (skin), neurology (nerves), ophthalmology (eyes), dietetics (nutrition), occupational therapy, and clinical psychology. These specialists will liaise with your local teams. If your child is unwell or requires further monitoring or intervention, you will be referred back to your local team. You may be asked to attend the multidisciplinary clinic at St Thomas’ sooner than your usual yearly appointment.
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