

## A simple template that can be used to develop tailored condition-specific information

### Name of condition/alternative name(s) of condition

If you are reading this information, then you and those around you are probably at the start of your journey with [condition]. You will find information here to help you understand [condition]. It might feel overwhelming at first, so take your time, write down any questions, and come back to find out more when you're ready.

Being tested for or diagnosed with a rare genetic condition can feel isolating, but please know that you are not alone. There are charities and support groups who can help you to navigate this journey and connect with other people and families living with [condition] who will be with you every step of the way.

### Overview

50-100 words providing a short overview about the condition.

### Symptoms

Every person is unique, and different people with the same condition can be affected differently. Some people may have every symptom, and some people will just have one or two symptoms. The most common symptoms are listed first.

- Bullet point list of symptoms.
- If there are lots of symptoms, put the most common symptoms first.
- If there are symptoms like rashes or lesions, mention that these may look different on different skin tones, or be harder to spot on darker skin.

### What causes [condition]?

Review and edit the text below and delete the sections that are not relevant for this condition. Keep any information about genetics very simple: sample text is suggested below to give you an understanding of the topline detail that is needed.

[Condition] is a genetic condition. Genes are the instruction manual for a person's body. We have more than 20,000 different genes that tell the body how to grow and do different things – from determining hair and eye colour to how our many organs work.

In people with genetic conditions, one or more of their genes don't instruct the body as we would expect, which can lead to changes in how their body works.

Every person has two copies of each gene, one from their mum and one from their dad. Genetic conditions can be caused by changes in either one or both copies. [Condition] is a dominant/recessive condition.

Dominant conditions are caused by changes to one copy of a gene. Recessive conditions are caused by changes to both copies of a gene.

Genetic conditions can be divided into three different categories: single gene, chromosomal or complex disorders. [Condition] is a single gene disorder / chromosomal disorder / complex disorder.

Single gene disorders are caused by changes (variants) in one particular gene.

Chromosomal disorders result from a change in the number of chromosomes, a missing or extra piece of a chromosome, or one or more chromosomes being arranged in a different way.

Complex disorders (also known as multifactorial or polygenic) are caused by changes to many different genes, often in complex interaction with environmental factors such as diet.

Say if something is known about a higher prevalence of the condition in any particular groups, for example, a particular biological sex or an ethnic group. This can reassure people that they are not alone and can explain why certain external organisations are being signposted.

## Diagnosis

Aim for 250 words maximum.

Include a couple of paragraphs describing:

- How the condition is usually diagnosed
- Any conventional testing or screening that may need to take place to confirm diagnosis.
- Any genetic testing that may be needed. If this information is likely to be shared with people prior to genetic testing for their condition, rather than after, you may want to explain how to access genomic testing, what happens at a genetic testing appointment, whether a family history or genetic tests for family members will be needed, and how long it takes to get the results. Emphasise that genetic tests are available from the NHS, and that genetic counselling is available before and after testing to help people make informed choices.
- For any invasive tests, highlight that chaperones or female doctors can be requested.

## Managing [condition]

Aim for **250** words maximum.

Consider whether management is the same for children and adults. If not, you may wish to have two sections answering these questions: one for children, and one for adults. Also consider if management is the same for boys and girls, particularly for X linked disorders, if not consider having two sections: one for boys and one for girls. You may also want a paragraph setting out what should be expected at the time of transition from children's services to adult's services.

Set out:

- how the condition is usually managed
- which part(s) of the NHS they are most likely to receive ongoing support from for their condition
- which healthcare professionals are likely to be involved
- which clinics are most likely to be visited, and whether they will offer face-to-face or virtual (telephone/video) options for different types of appointment
- what support might be put in place (for example, from speech and language therapists, physiotherapists, learning disability nurses and support groups)
- whether there are lifestyle changes such as diet that can help manage the condition (and remember that cultural sensitivity may be needed here)
- that people can take an advocate or family member to appointments to support them if needed and ask the NHS to provide a translator if this would help them to understand the information provided at appointments.

## Treating [condition]

Aim for 250 words maximum.

Are there any treatments available from the NHS, either for particular symptoms or for the condition as a whole? If so, add a short paragraph about each treatment here. If different treatments are recommended for use by the NHS in England, Scotland, Wales and Northern Ireland, you may wish to highlight this too.

If there are potential treatments that are currently going through clinical trials or available through compassionate use schemes, you can mention them to give people hope for the future. However, you should make clear that they are not yet licensed for use in the UK or widely available through the NHS.

## Will other people in my family be affected by [condition]?

A clinical geneticist or genetic counsellor will be able to provide specific advice about how [condition] might affect other members of your family.

If you are planning to have children in future, you may want to seek advice from your GP or hospital consultant on what the chances are of having another child with this condition, and if so, what reproductive choices are available to you such as preimplantation genetic testing. You can ask for counselling before and after prenatal testing to help you think through your options.

### Further information

List – signpost to more detailed information resources produced by charities and support groups from the UK and overseas here. Consider the audience for this document when deciding whether it is also appropriate to signpost to clinical or research materials about [condition].

### Additional support

List – signpost to condition-specific charities and support groups that can provide information and peer to peer support.

Add date doc created / date for review

About [Name of charity]

Add a short paragraph about your charity or support group and a link to your website. Add a line stating 'If you have any comments or questions about this information, please contact [email address].

Disclaimer

We have tried to make sure that this information is correct. We do not accept liability for any mistakes or any information not included. For up to date information about medicines and treatments you should talk to your doctor. Copyright © YEAR [Name of charity].