PATIENT INFORMATION LEAFLET

# CYLD CUTANEOUS SYNDROME

## WHAT ARE THE AIMS OF THIS LEAFLET?

This leaflet has been written to help you understand more about CYLD cutaneous syndrome (CCS). It tells you what it is, what causes it and how it can be treated. It also explains where you can find out more about it. CCS has previously been known as:

- Brooke-Spiegler syndrome,
- Familial cylindromatosis,
- Multiple familial trichoepitheliomas.

Recent research supports CYLD cutaneous syndrome as the most appropriate name for this condition. This is because the older names did not include the right information needed to help affected families get the right support.

## WHAT IS CYLD CUTANEOUS SYNDROME?

CCS is a rare genetic condition where patients develop multiple benign (non-cancerous) skin tumours (an abnormal growth of body tissue) on the scalp, face and body. These include:

- cylindromas – they are usually painless and present as pink lumps on the scalp and torso. In black or brown skin, they can appear darker than the surrounding skin.
- spiradenomas – they are often painful and can have a blue or black colour. In black or brown skin, they can appear darker than the surrounding skin.
- trichoepitheliomas – they are skin-coloured small lumps often seen on the skin around the nose. In black or brown skin, they are the same colour as the surrounding skin.

Features of cylindroma and spiradenoma can often be found in a single skin lump in patients with CCS.

## WHAT CAUSES CYLD CUTANEOUS SYNDROME?

CCS occurs due to a variation in a piece of DNA called the *CYLD* gene (pronounced "SILD").

## DOES CYLD CUTANEOUS SYNDROME RUN IN FAMILIES?

CCS patients usually have a family history, but some people may develop it for the first time in their family.

In patients with multiple CCS skin tumours, these are hereditary and there is a 50% (1 in 2) chance of passing the condition on to their children.

A blood test can be carried out to determine whether someone has the variation in the *CYLD* gene. Your dermatologist can advise you about the blood tests that are available to confirm a potential genetic link.

## WHAT DOES CYLD CUTANEOUS SYNDROME LOOK LIKE?

CCS skin tumours are benign but rarely there may become malignant

- What are they made of? – They are made from the same cells that are seen in hair follicles and sweat glands in the skin.
- Why do some hurt and others do not ?– Some benign skin tumours can hurt but it is not understood why. Research is being done to try to understand this.
- Where can they grow? – CCS skin tumours can be found all over the body. They are particularly common in hairy parts of the body such as the face, scalp, chest, stomach and pubic area. They can develop on the skin overlying breast tissue. If you are attending for a mammogram, it is helpful to inform screening staff about this condition. This is because they may see

CCS skin tumours on a mammogram. A biopsy may be necessary if there is uncertainty following a mammogram. A biopsy involves taking a small sample of the tumour to be looked at in a laboratory to see what type of cells it is made from.

- How big do they get? The majority are small, no bigger than the size of a grape (1-2 cm). Some can grow to be 5-10 cm across, but this is uncommon.

## WHAT PROBLEMS CAN THEY CAUSE?

- These skin tumours increase in number over time and can be painful.
- The skin over a benign tumour can break down and form an ulcer, which can weep and bleed. This requires medical attention.
- CCS skin tumours in the ear canal can result in deafness. This is due to the tumour blocking the ear canal.
- In a small number of patients (about 5%), a tumour can develop in the salivary glands. These are usually benign and can be treated with surgery. If you suspect a lump in the skin in front of the ear or below the jaw where the salivary glands are located, seek medical advice as you may need a biopsy to plan treatment.
- Rarely, some CCS skin tumours can become malignant or cancerous, and spread to other parts of the body such as the lungs or liver.

## HOW IS CYLD CUTANEOUS SYNDROME DIAGNOSED?

CYLD cutaneous syndrome is usually diagnosed by a dermatologist or a geneticist who are specialist doctors. This involves examining the skin, talking about your family history, and in some cases taking a surgical biopsy of a skin tumour in a hospital. Genetic testing can be offered where necessary to confirm a clinical diagnosis of CCS.

## CAN CYLD CUTANEOUS SYNDROME BE CURED?

There is currently no cure for CCS. People often develop multiple growths, throughout their life. Although the growths are harmless, uncommonly, a cancerous growth (also known as malignant tumour) may develop on the skin. Examples include cylindrocarcinoma, spiradenocarcinoma, and basal cell carcinoma. There may also be a higher risk of internal cancers like saliva gland cancers.

However, there are treatments that can help control CCS.

## HOW CAN CYLD CUTANEOUS SYNDROME BE TREATED?

- What treatment options are available? – Different removal techniques may be offered depending on the type of tumour and location on the body. This can be done by various types of surgery, laser, or cryotherapy, or a combination of these methods. Your dermatologist can discuss which methods will suit you.
- Is it dangerous to leave them untreated? – Ideally, growing tumours should be treated at an early stage. This is to minimise surgery later on and lower the risk of future problems. It is important for patients to self-examine and tell the doctor about CCS tumours that grow rapidly. Also tell your doctor if you develop an ulcer or bleed from the overlying skin. If you are concerned about a tumour, seek medical advice.
- Can benign skin tumours come back after an operation? – Yes, benign tumours can grow back where the scar is, after removal, as it may not be possible to always remove all of the tumour cells. New benign tumours can also grow nearby from other hairs.

## FREQUENTLY ASKED QUESTIONS

### *Where can I get more information about CYLD cutaneous syndrome?*

Online resources informed by clinical research studies of CCS patients are available at:

- Gene Reviews – CYLD cutaneous syndrome - National Center for Biotechnology Information (NCBI)

  www.ncbi.nlm.nih.gov/books/NBK555820/

- Genetic and Rare Diseases Information Center (GARD) rarediseases.info.nih.gov/diseases/10179/brooke-spiegler-syndrome
- Medline Plus medlineplus.gov/genetics/condition/cyld-cutaneous-syndrome/
- Orphanet www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=79493

### *Links to patient support groups*

www.facebook.com/groups - Multiple Cylindromas Support Group

These questions and answers are meant to act as guidance for patients who have been diagnosed by a dermatologist or geneticist. It is not meant to replace regular clinical care and advice. If you have any questions, please contact your general practitioner or dermatologist.

*Please note that the British Association of Dermatologists (BAD) provides web links to additional resources to help people access a range of information about their treatment or skin condition. The views expressed in these external resources may not be shared by the BAD or its members. The BAD has no control of and does not endorse the content of external links.*

This leaflet aims to provide accurate information about the subject and is a consensus of the views held by representatives of the British Association of Dermatologists: individual patient circumstances may differ, which might alter both the advice and course of therapy given to you by your doctor.

*This leaflet has been assessed for readability by the British Association of Dermatologists' Patient Information Lay Review Panel*

**BRITISH ASSOCIATION OF DERMATOLOGISTS**

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PRODUCED | JANUARY 2021
UPDATED | MARCH 2026
NEXT REVIEW DATE | MARCH 2029