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, what can be done about it, and where you can find out more about it. CCS has previously been referred to by a range of names, including Brooke-Spiegler syndrome, “Familial cylindromatosis” and “Multiple familial trichoepitheliomas”. Recent research supports CYLD cutaneous syndrome as the most appropriate name for this condition, as none of the older names are useful for predicting disease prognosis or informing genetic counselling in an affected family.

What is CYLD cutaneous syndrome?

CCS is a rare genetic condition which causes patients to develop multiple benign skin tumours on the scalp and body. The benign skin tumours may be called cylindromas, spiradenomas or trichoepitheliomas, and some patients may have a combination of these tumours. Cylindromas are benign skin tumours that arise from hair follicles, which present as pink lumps on the scalp and trunk. Spiradenomas are benign skin tumours that are often painful, and can have a blue or black colour, and are related to cylindroma. Features of cylindroma and spiradenoma can often be found in a single skin lump in patients with CCS. Trichoepithelioma are skin-coloured small lumps often seen on the skin around the nose.

What causes CYLD cutaneous syndrome?

- CCS occurs due to a fault in a piece of DNA called the *CYLD* gene (pronounced “SILD”).

Is CYLD cutaneous syndrome hereditary?

- In patients with multiple CCS skin tumours, these are hereditary and are typically passed on to half of their children. There is usually a family history, but some people may develop it for the first time in their family. A blood test can be carried out to determine whether or not someone has the

fault in the *CYLD* gene. Your clinician can advise you on blood tests that are available to confirm a potential genetic link.

What does CYLD cutaneous syndrome look like?

CCS skin tumours are usually benign, which means they are not cancerous.

- **What are they made of?** – They are made of the same cells that are seen in hair follicles and sweat glands in the skin.
- **Why do some hurt and others don't?** – 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?** – Skin tumours can be found throughout the body surface, with a preference for hairy parts of the body. They are most common on the face and scalp, trunk and pubic area. They can develop on the skin overlying breast tissue, and it is helpful to inform screening staff if attending for a mammogram about this condition as they may see CCS skin tumours on a mammogram. A biopsy may be necessary if there is uncertainty following a mammogram.
- **How big do they get?** The majority are small, no bigger than the size of a grape (2-3 cm), but some can grow to be 5-10 cm across, though this is uncommon.

What problems can they cause?

- These skin tumours grow progressively and can be painful.
- The skin over a benign tumour can break down and form an ulcer, which can weep and bleed.
- Cylindromas in the ear canal can result in deafness due to the tumour blocking the ear canal.
- In a minority of patients (about 5%), a tumour can develop in the salivary gland. These can be treated with surgery. Seek medical advice if you suspect a lump in the skin in front of the ear or below the jaw where the salivary glands are located.
- Uncommonly, some tumours can become cancerous, and spread to other parts of the body such as the lung or liver.

How is CYLD cutaneous syndrome diagnosed?

CYLD cutaneous syndrome is usually diagnosed by a dermatologist or a geneticist. This involves a skin examination, a family history, and in some cases a surgical biopsy of a skin tumour in a hospital. Genetic testing is available, and can inform genetic counselling.

Can CYLD cutaneous syndrome be cured?

No, this is a genetic condition and there is currently no cure. However, there are treatments that can help control CCS.

How can CYLD cutaneous syndrome be treated?

- **What treatment options are available?** – Different surgical techniques have been developed and are tailored to the type of tumour and location on the body. Given the rare nature of this condition, this should be discussed with your dermatologist.
- **Is it dangerous to leave them untreated?** – Ideally, growing tumours should be treated at an early stage to minimise more extensive surgery later on. It is important for patients to self-examine, and report tumours that grow rapidly, develop an ulcer in the overlying skin or bleed to their doctor for assessment. If you are concerned about a tumour, seek medical advice.
- **Can benign skin tumours come back after an operation?** – Yes, benign tumours can grow at the scar site or new benign tumours can grow nearby.

Frequently Asked Questions

Where can I get more information about CYLD cutaneous syndrome?

Web links to patient support groups

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

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

- National Center for Biotechnology Information (NCBI) https://www.ncbi.nlm.nih.gov/books/NBK555820/
- Genetic and Rare Diseases Information Center (GARD) https://rarediseases.info.nih.gov/diseases/10179/brooke-spiegler-syndrome
- Medline Plus https://medlineplus.gov/genetics/condition/cyld-cutaneous-syndrome/
- Orphanet https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=79493

These questions and answers are meant to act as guidance for patients who have been diagnosed by a dermatologist. It is not meant to replace regular

clinical care and advice. If you have any questions, please contact your general practitioner or dermatologist.

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

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