

PE2080/D: Implement the recommended screening guidelines for people with Li Fraumeni Syndrome

Cancer Research UK written submission, 19 June 2024

Thank you for your letter of 17th May 2024 on behalf of the Citizen Participation and Public Petitions Committee regarding screening of Li Fraumeni Syndrome.

Li Fraumeni Syndrome (LFS) is a genetic condition that increases people's risk of certain childhood and adult cancers. It's extremely rare, so data is limited, but [around 80% of people living with LFS will develop cancer by age 70.](#)

[Researchers estimate that there are a few thousand people in the UK with LFS, though only around 500 to 700 people have received a diagnosis.](#) Almost all of them are close to someone else with the condition. Many will have lost someone they love because of it.

The UK Cancer Genetics Group (UKCGG) is a Special Interest Group of the British Society of Genetic Medicine (BSGM). UKCGG is a national, multi-disciplinary organisation with membership including clinicians, genetic counsellors and scientists with an interest in the hereditary aspects of cancer. In 2020, [the UKCGG published guidelines for the management of patients with LFS](#), which included recommendations for a UK-wide surveillance protocol.

We would encourage the Scottish Government to engage in further dialogue with the UK National Screening Committee (UK NSC), an independent expert advisory group that advises all four UK nations on new and existing screening programmes, who have not at present made any recommendations about targeted surveillance or screening programmes for those with Li Fraumeni Syndrome. We would also advise the Scottish Government to strengthen links with the research community as there are a number of clinical trials currently investigating LFS (see details below) so that consensus can be achieved for the benefit of all Scottish patients.

Researchers are developing ways to either prevent cancer or detect it sooner in people with LFS, who are often excluded from larger prevention studies. Their cancer risk levels are so much higher than the general population that they can skew the results. [Cancer Research UK is funding the first of a new kind of precision cancer prevention trial, which is looking at whether an anti-diabetic drug called metformin works to reduce cancer risk in people with LFS.](#) [The ATLAS study](#), co-funded by Cancer Research UK, aims to detect cancers earlier in people with LFS using a blood test, which could be used as a supplementary screening tool, in conjunction with MRI scanning.

[The SIGNIFIED trial, funded by the Small Business Research Initiative \(SBRI\), monitors patients with LFS patients with two whole-body MRI scans \(WBMRI\) annually, aiming to detect cancer at its earliest stages.](#) [This study builds on the previous Cancer Research UK funded SIGNIFY trial](#), which concluded that WBMRI scans could help diagnose people with LFS and might be a useful screening test for this group.

I would like to thank you again for raising this petition with Cancer Research UK and I hope that you find this response helpful. Please do not hesitate to contact me further if you would like any more information on any of the work Cancer Research UK is engaged in regarding equity in screening.