Citizen Participation and Public Petitions Committee
Wednesday 10 December 2025
19th Meeting, 2025 (Session 6)

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

Introduction

Petitioner	Louise Mckendrick
Petition summary	Calling on the Scottish Parliament to urge the Scottish Government to implement screening for people with, or at risk of, Li Fraumeni Syndrome (TP53 mutation), in line with the guidelines recommended by the UK Cancer Genetics Group.
Webpage	https://petitions.parliament.scot/petitions/PE2080

1. The Committee last considered this petition at its meeting on 15 May 2024. At that meeting, the Committee agreed to write to Cancer Research UK, the Scottish Government and NHS Scotland genetic laboratories in Aberdeen, Dundee, Edinburgh and Glasgow.

2. The petition summary is included in **Annexe A** and the Official Report of the Committee's last consideration of this petition is at **Annexe B**.

3. The Committee has received new written submissions from the Scottish Government, the Scottish Hereditary Cancer Genetics Group and Cancer Research UK, which are set out in **Annexe C**.

4. Written submissions received prior to the Committee's last consideration can be found on the petition's webpage.

5. Further background information about this petition can be found in the SPICe briefing for this petition.

6. The Scottish Government gave its initial response to the petition on 14 February 2024.

7. Every petition collects signatures while it remains under consideration. At the time of writing, 1,033 signatures have been received on this petition.

8. At its meeting on 24 September 2025, the Committee took evidence on thematic healthcare issues that have been raised in multiple petitions, including this petition.

Action

9. The Committee is invited to consider what action it wishes to take.

Clerks to the Committee
December 2025

Annexe A: Summary of petition

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

Petitioner

Louise Mckendrick

Date Lodged

22 January 2024

Petition summary

Calling on the Scottish Parliament to urge the Scottish Government to implement screening for people with, or at risk of, Li Fraumeni Syndrome (TP53 mutation), in line with the guidelines recommended by the UK Cancer Genetics Group.

Previous action

I have made a complaint to NHS Greater Glasgow and Clyde.

I have also recently contacted my local MSP and my MP for East Renfrewshire.

Background information

Li Fraumeni TP53 mutation is a genetic syndrome that predisposes a person to cancer, usually of an aggressive type.

The UK Cancer Genetics Group (UKCGG) have made recommendations for screening which have been implemented in parts of England, however they have not been implemented in Scotland. I am calling on the Scottish Government to implement screening for Li Fraumeni patients in line with the guidelines set out in the **UKCGG paper in the Journal of Medical Genetics**.

Annexe B: Extract from Official Report of last consideration of PE2080 on 15 May 2024

The Convener: PE2080, which was lodged by Louise McKendrick, calls on the Scottish Parliament to urge the Scottish Government to implement screening for people with, or at risk of, Li-Fraumeni syndrome due to TP53 mutation, in line with the guidelines recommended by the UK Cancer Genetics Group. LFS, as it is known, is rare, with researchers estimating that a few thousand people in the UK have it. The UK Cancer Genetics Group guidelines recommend regular screening for people with LFS.

The SPICe briefing that we have received notes that the guidelines that are cited by the petitioner do not actually recommend routine cancer screening for those who are identified as being at risk of having the TP53 gene mutation that causes LFS. Instead, the guidelines recommend that they be offered appropriate counselling and support and encouraged to consider whether they want to be tested for the TP53 gene variant.

The Scottish Government's response to the petition states:

"The UK Cancer Genetics Group ... guidelines are endorsed and supported by clinical genetics teams ... across Scotland."

However, it adds:

"We are aware of variation in how these guidelines are implemented across ... health boards in Scotland and of acute demand for MRI procedures in particular."

The Government is

"considering how to better signpost management guidelines and ... improve the consistency in implementation".

In view of the Government's position and the UK Cancer Genetics Group guidelines, do members have comments or suggestions for action?

Fergus Ewing: Reading the background information on the petition, I note that the LFS TP53 mutation is

"a genetic syndrome that predisposes a person to cancer, usually of an aggressive type"

and that the relevant authoritative body—the UK Cancer Genetics Group—has

"made recommendations for screening which have been implemented in parts of England, however they have not been implemented in Scotland."

I see that there is a reasonably sympathetic reply from the chief operating officer of NHS Scotland, although it does not really give much information, other than saying that there are good intentions all round but that there is pressure on MRI scans, which is understandable.

In light of that, I certainly do not think that we should close the petition. We need to get more information. I would like us to go back to the Scottish Government and ask whether it can provide more information on what services are available in each health board. Given that this group of people are predisposed to cancer of an aggressive type, it seems to me to be a very serious disease, albeit a rare one. I see that the petition has attracted nearly 1,000 signatures, so there is obviously considerable concern. We should ask the Scottish Government to provide that further information in light of the gravity of the matter. Secondly, I suggest that we write to Cancer Research UK to seek its views on the action that is called for in the petition.

I wonder whether there are other things that we might do, convener. Maybe colleagues have other thoughts. It does not seem to me that the current response is adequate.

The Convener: Do colleagues have any suggestions that we might add to Mr Ewing's?

David Torrance: We should write to NHS Scotland's genetic laboratories in Aberdeen, Dundee, Edinburgh and Glasgow to seek further detail on the work that they do to support the implementation of the UK Cancer Genetics Group guidelines and information on the genetic testing and support that is available for those who are at risk of having a faulty TP53 gene.

The Convener: We have a series of suggestions on how we should proceed. Are members content that we proceed on that basis?

Members *indicated agreement.*

The Convener: We thank the petitioner for raising this fresh issue with us. We note the number of signatures that the petition has attracted. We will seek to take the issue further forward.

Annexe C: Written submissions

Scottish Government written submission, 10 June 2024

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

Thank you for your letter of 17 May 2024 on behalf of the Citizen Participation and Public Petitions Committee in relation to PE2080, regarding screening around Li-Fraumeni Syndrome (TP53 mutation).

Background

Li-Fraumeni syndrome is a rare hereditary genetic condition caused by changes in the TP53 gene. This gene would normally be protective against cancer by helping to control the growth of cells within the body. Anyone in Scotland with a family history of specific cancers can be referred for a genomic test at an accredited NHS genomic laboratory to identify if they are at risk of developing Li-Fraumeni syndrome. People diagnosed with Li-Fraumeni syndrome are managed by clinical genetic services, with screening procedures carried out across the 14 territorial health boards.

Action called for in the petition

The Committee requested further information on the services that are, or should be, available in each health board for people with Li Fraumeni Syndrome to support the implementation of the UK Cancer Genetic Group guidelines.

The Scottish Government is pleased to offer the Committee our views on the follow-up action:

The UK Cancer Genetics Group (UKCGG) guidelines are endorsed and supported by clinical genetics teams working across Scotland.

As such, people diagnosed with Li Fraumeni Syndrome (with TP53 mutation) should be offered a protocol of screening procedures subject to eligibility criteria set out within the UKCGG guidance. The recommended screening protocol will vary depending on the results of the genetic tests.

As per our previous response we are aware of variation in how these guidelines are implemented across the 14 territorial health boards in Scotland and of acute demand for MRI procedures in particular. We are working to better signpost management guidelines and support resources to improve the consistency in implementation across Scotland.

Screening in Scotland is guided by the recommendations of the UK National Screening Committee (UK NSC), an independent expert advisory group that advises all four UK nations on new and existing screening programmes. They will only make a recommendation to implement national screening programmes after assessing the potential benefit and harms of screening the population and such recommendations are based on evidence that the harms outweigh the benefits. At present, the UK

NSC has not made any recommendations about targeted screening programmes for those with Li-Fraumeni Syndrome.

I would like to thank you again for raising this concern with the Scottish Government and I hope that you find this response helpful.

Directorate for Chief Operating Officer, NHS Scotland

Scottish Hereditary Cancer Genetics Group written submission, 13 June 2024

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

As the petitioner has rightly pointed out, UK consensus guidelines for cancer surveillance for individuals with Li Fraumeni syndrome were published by the UK Cancer Genetics Group in 2020. The guidance recommends annual whole-body MRI from birth which takes 90-120 minutes to complete the scan in adults. In addition, there is considerable radiologist time required in reporting each scan (takes approximately three times the length of time compared to conventional MRI and require to be double read effectively multiplying the radiologists' time for these scans by a factor of 6). Local health boards have therefore found this very difficult to implement without any additional resource.

NHS Grampian, NHS Tayside, NHS Lothian, and NHS Fife are all offering surveillance in patients with Li Fraumeni syndrome in line with these guidelines (despite the considerable strain on services) but given the larger numbers of affected patients in the West of Scotland, they have not yet been able to implement these guidelines. The Scottish Hereditary Cancer Genetics Group has raised this issue on several occasions, initially with a submission to Health Improvement Scotland (informed not within their remit) and subsequently with the Scottish Cancer and Genomic Policy Teams. This is just one of several issues that were recently presented to the above policy teams concerning management of patients at high risk of cancer which requires a national consensus and approach to service delivery with adequate funding. There is a clear gap in Scotland regarding a national process for managing these patients and this is becoming increasingly problematic with the advancements in precision cancer treatments and management guidelines around surveillance in people at increased risk. Although the need and potential benefits in this area were highlighted and recognised, there has been no move to address these issues which continues to rely on local efforts of individual clinicians and good luck!

Cancer Research UK written submission, 19 June 2024

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

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.