

# Scottish Government submission of 14 February 2024

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

Thank you for your email of 22 January 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 Scottish Government is pleased to offer the Committee our views on the following action called for in the petition:

- ***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***

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

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. Therefore, we are considering how to better signpost management guidelines and support resources to improve the consistency in implementation across Scotland. We will

continue to work with Boards and clinical communities to ensure that the requirements of people with Li-Fraumeni syndrome are understood.

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**