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

Scottish Government written submission, 10 June 2024

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