Inquiry into the equitable access to diagnosis and treatment for individuals with rare and less common cancers, including neuroendocrine cancer

Sydney Hearing –31 January 2024

Question on Notice from Senator Wendy Askew

Rare Cancers Australia

1. Li-Fraumeni Syndrome statistics

Li-Fraumeni syndrome (LFS) is a cancer predisposition syndrome associated with high risks for a diverse spectrum of childhood- and adult-onset malignancies. The lifetime risk of cancer in individuals with LFS is ≥70% for men and ≥90% for women. https://www.ncbi.nlm.nih.gov/books/NBK1311/#:~:text=Li%2DFraumeni%20syndrome%20(LFS),and%20%E2%89%A590%25%20for%20women.

Individuals with LFS have an approximately 50% chance of developing cancer by age 40, and up to a 90% percent chance by age 60. Females with LFS have a risk as high as a 90% risk of developing cancer in their lifetime due to their markedly increased risk of breast cancer. Many individuals with LFS develop two or more primary cancers over their lifetime which generally occur in age-related phases. https://www.lfsassociation.org/what-is-lfs/

2. The use of MRI's in screening and early detection of cancer for familial syndromes

Further information regarding the use of MRI's in screening and early detection of cancer for familial syndromes.

Studies have been conducted to screen and detect cancers in the population with the genetic mutation TP53, this population has a significantly higher risk of developing cancer than the general population. TP53 can also be diagnosed as Le Fraumeni Syndrome (LFS). Following successful studies, a Medicare number has been issues for use of scheduled MRI scanning in families with LFS.

The original funding was from a MRFF research grant to the Genomic Cancer Medicines Program (Clinical Investigator – A/Prof Mandy Ballinger). Protocol name: Surveillance study Multi-Organ Cancer prone syndromes (SMOC). Subsequently there is on-going screening for similar multi-organ syndromes with high risk of cancer in children, protocol: SMOC junior. This work is being funded by MRFF funding and philanthropic funding.

Current objective is to secure further research funding to continue to build evidence to establish screening protocols for high-risk populations, and ultimately translate that evidence to clinical practice.