



## Service Priorities and Programmes Electronic Presentations

**Convention ID:** 1142

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### **Pilot Screening and Monitoring Program for Patients with Cancer Predisposing Li Fraumeni Syndrome in Prince of Wales Hospital, Hong Kong**

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#### **Keywords:**

Cancer

Li Fraumeni Syndrome

Surveillance

#### **Introduction**

Li Fraumeni syndrome (LFS) is a cancer predisposing syndrome due to mutation of tumor suppressor gene TP53. Test for TP53 mutation was initially offered to selected paediatric oncology patients and families in Prince of Wales Hospital. This was followed by setting up of a pilot multidisciplinary clinic (familial-cancer clinic) comprised of paediatricians, clinical oncologists, surgeons, pathologists and radiologists in 2014.

#### **Objectives**

Provide counselling and cancer surveillance to LFS patients.

#### **Methodology**

Since 2014, patients with TP53 mutation were followed up in the multidisciplinary clinic. They were given advice on healthy life-style, signs and symptoms of cancer and breast-examination in females. Cancer surveillance for these patients included annual physical examination, blood tests, radiological assessments (which consisted of annual magnetic resonance imaging of brain, half-yearly ultrasonography of abdomen and pelvis, ultrasonography of breast (female patients only)) and upper and lower gastrointestinal endoscopy at 3 to 5 years intervals.

#### **Result**

There were 8 subjects with childhood cancer identified to be carrier of TP53 mutation. Indications for screening for mutation were as follows: history of 2 or more cancers (N=4, 50%), history of cancer with strong association with TP53 mutation (N=3, 37.5%), strong family history of cancer (N=1, 12.5%). 2 patients died from progressive disease of the first malignancy. Subsequently, 6 of their family members were found to have the mutation, 5 being asymptomatic for cancer while 1 had history of 2 cancers. Five patients developed new cancers after the confirmation of TP53 mutation. An asymptomatic carrier was found to have early stage carcinoma of colon picked up by screening colonoscopy, allowing timely surgical resection. The others being new cancers in index patients and included carcinoma of stomach (Stage IIB, post partial gastrectomy for 2 years), ductal carcinoma of breast (Stage IIA) detected by screening USG of breast and awaiting surgery, squamous cell carcinoma over eyebrow (Stage 1 with complete resection) and carcinoma of lung (Stage 4). With increase in patient and physician awareness of cancer risk associated with LFS, early identification of cancers and timely intervention might be feasible. A systemic approach to screen for high risk patients for the mutation and monitoring of affected individuals is of clinical importance.