Patients with genetic tumour risk syndromes (genturis) are at high risk of developing tumours in multiple organ systems at a young age.

Early identification and diagnosis are the critical factors to improve prognosis and access to appropriate and timely management.

There are > 40 different genturis, including TP53-related cancer syndrome (hTP53rc, Li-Fraumeni syndrome) and PTEN hamartoma tumour syndrome (PHTS, Cowden syndrome).

### Method

- We established two separate multidisciplinary teams of experts representing all relevant disciplines and patients’ representatives.
- 337 (hTP53rc) and 131 (PHTS) published articles were assessed in the guideline development.
- The guidelines were based on the best evidence and expert consensus.
- A lay-persons’ version and a ‘pocket guide’ was also developed.

### Surveillance Protocol for Heritable TP53-related cancer syndrome (hTP53rc)

#### Recommendations

- Clinical examination and abdominal ultrasound biannually.
- Where individuals harbour a TP53-pathogenic variant, known to be associated with childhood cancers, annual whole-body-MRI and brain-MRI from the first year of life is recommended.

In adults (>18), the surveillance protocol includes:

- Annual clinical examination.
- Annual whole-body-MRI.
- Annual breast-MRI in females from 20 until 65 years.
- Annual brain-MRI until 50 years.

### Cancer Surveillance Protocol for individuals with PTEN hamartoma tumour syndrome (PHTS)

#### Recommendations

- Breast cancer annual MRI from age 30.
- Thyroid cancer annual ultrasound from age 18.
- Renal cancer ultrasound two-yearly, from age 40.
- Colorectal cancer baseline colonoscopy at age 35-40.
- Melanoma baseline skin examination at age 30.