Guidelines for management of tumour risk in PTEN hamartoma syndrome
(UK Cancer Genetic Group – May 2017)

Cancer risk	Risk data	Screening advice
Breast	Cancer – lifetime up to 85% Average age at diagnosis 38-46 High incidence of fibrocystic breast disease	*BRCA*-equivalent (annual MRI from age 30, mammography from 40)
Thyroid	Cancer – lifetime 35% (usually follicular, rarely papillary, never medullary) Median age at diagnosis 37 Up to 75% risk of multinodular goitre, adenomatous nodules & follicular adenomas	As a minimum annual screen from 16 by USS. Younger as guided by family history or after informed discussion with family.
Endometrial	Cancer – lifetime up to 28% Risk starts late 30s – early 40s Benign uterine fibroids very common.	Refer to specialist Gynaecologist age 35-40 for discussion regarding screening options and management of non-cancer manifestations Consider risk-reducing hysterectomy
Renal	Cancer – lifetime up to 35% (mostly papillary) Risk starts late 40s	Annual renal USS/MRI from 40
Colorectal	Cancer – lifetime up to 9% Risk starts late 30s More than 90% have polyps, which may be symptomatic	Ascertainment colonoscopy at age 35 and 55 Polyp f/u as required
Skin & vascular system	Melanoma – ~5% Many non-malignant lesions	Baseline dermatological review & appropriate f/u
Brain	Lhermitte-Duclos disease – up to 32%	Brain MRI only if symptomatic

V1.0 - 4 MAY 2017

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