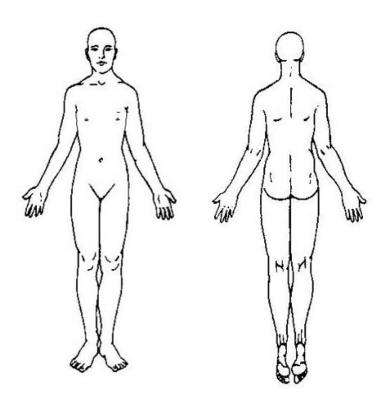
| MEDICAL GENOMICS LABORATORY: PTEN PHENOTYPIC CHECKLIST FORM | |
|---|--|
| | |
| Patient ID:Referring Physician | Date of Evam (MM/DD/YY)// |
| Referring Physician | |
| DEMOGRAPHIC INFORMATION | |
| Gender: ☐ Male ☐ Female | |
| Ethnicity: Mother - | |
| CLINICAL DIAGNOSIS | |
| | ☐ Bannayan-Riley-Ruvalcaba syndrome (BRRS) |
| | □ Proteus-like syndrome |
| ☐ Macrocephaly/Autism syndrome | □ Other |
| FARMLY LICTORY | |
| FAMILY HISTORY | □ Halanasan |
| ☐ Sporadic ☐ Familial | Unknown |
| Familial cases: Please provide pedigree and details on the affection status of family members on a separate page. | |
| General Information | |
| | |
| Weight kg or lb Height cm or ft Head circumference cm | |
| CNS | <u>Breasts</u> |
| ☐ Macrocephaly (HC > 98th percentile) | ☐ Fibrocystic disease |
| ☐ Mental retardation/Developmental delay | ☐ Breast cancer (type) |
| IQ | BRCA testing? □ No □ Yes |
| ☐ Autism/ pervasive developmental disorder | If yes, was mutation identified? |
| ☐ Lehrmitte-Duclos disease (LDD) | ☐ Yes ☐ No Thyroid |
| ☐ Other CNS tumors | ☐ Thyroid cancer (non-medullary) |
| Skin/Mucosa | ☐ Benign multinodular goiter |
| ☐ Facial trichilemmomas | ☐ Adenomatous nodules and follicular adenomas |
| □ 0-2 lesions □ ≥ 3 lesions | ☐ Other thyroid lesions |
| | Gastrointestinal tract |
| ☐ Cutaneous facial papules | ☐ Hamartomatous intestinal polyps |
| ☐ Oral mucosal papillomatosis | ☐ GI malignancy (specify) Genitourinary |
| ☐ Acral keratosis | Benign uterine fibroids |
| ☐ 0-5 lesions ☐ ≥ 6 lesions | ☐ Endometrial cancer |
| Lipomas | ☐ Renal cell cancer |
| ☐ Regional absence of fat | ☐ Bilateral ovarian cystadenomas |
| ☐ Fibromas | \square Genitourinary malformation (specify) |
| ☐ Pigmented macules of the glans penis | Disproportionate overgrowth |
| ☐ Connective tissue nevi | ☐ Limbs (arms/legs, hands/feet/digits) |
| ☐ Epidermal nevus | ☐ Skull (hyperostoses)☐ Vertebrae (megaspondylodysplasia) |
| ☐ Vascular malformations | ☐ Viscera (spleen/thymus) |
| ☐ Capillary ☐ Venous ☐ Lymphatic | Others |
| ☐ Other skin lesions | ☐ Parotid monomorphic adenoma |
| | ☐ Facial dysmorphism (specify) |

** Please draw location/size of cutaneous lesions



Family pedigree – in familial cases

