

International Cowden Consortium operational diagnostic criteria (version 2000)

Pathognomonic criteria:

- Mucocutaneous lesions
 - Trichilemmomas (facial)
 - Acral keratoses
 - Papillomatous papules
 - Mucosal lesions

Major criteria:

- Breast cancer
- Thyroid cancer (especially follicular)
- Macrocephaly (>97th percentile)
- Endometrial carcinoma
- Lhermitte-Duclos disease (LDD), defined as presence of a cerebellar dysplastic gangliocytoma

Minor criteria:

- Other thyroid lesions (for example, goiter or nodule)
- Mental retardation
- Hamartomatous intestinal polyps
- Lipomas
- Fibrocystic breast disease
- Fibromas
- Genitourinary tumors (for example uterine fibroids, renal cell carcinoma) or malformations

Requirements for Diagnosis

- Mucocutaneous lesions if:
 - 6 or more facial papules (of which 3 or more must be trichilemmoma), or
 - Cutaneous facial papules and oral mucosal papillomatosis, or
 - Oral mucosal papillomatosis and acral keratoses, or
 - 6 or more palmoplantar keratoses
- Two or more major criteria met (one must be macrocephaly or LDD)
- One major criteria and three minor criteria
- Four minor criteria

For individuals with a family member diagnosed with CS

- A pathognomonic mucocutaneous lesion
- Any one major criteria with or without minor criteria
- Two minor criteria