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Cowden disease

Cowden disease is also known as 'Cowden's syndrome' and 'multiple hamartoma syndrome'.

What is Cowden disease?

Cowden disease is a rare inherited condition characterised by:

Cutaneous features

Cowden disease



Facial papule



Acral keratoses



Close-up

- Benign lesions of the skin and mucosa, known as hamartomas (benign tumours made up of a mixture of mature cells normally found in that tissue). At least one of the four types of skin lesions are present in nearly all cases.
 - Facial papules – flesh-coloured flat-topped dry or warty 1–5mm papules around the mouth, nostrils and eyes.
 - Oral lesions – numerous 1–3 mm smooth whitish spots on the gums and palate that join together to create a cobblestone appearance known as papillomatosis.
 - Acral keratoses – flesh-coloured or slightly pigmented smooth or warty papules on the upper surface of hands and feet. These occur in more than 60% of patients.
 - Palmoplantar keratoses – scaly spots on the palms and soles occur in about 40% of patients.
- Other skin lesions occurring less frequently include [lipomas](#), neuromas and [haemangiomas](#).

Non-cutaneous features

- Abnormalities of the thyroid are present in about 60% of patients. These are usually harmless growths but occasionally may be cancerous.
- Breast tumours: These are the most important non-cutaneous association. Fibrocystic disease resulting in benign lumps in the breasts is present in about 75% of women. Breast cancer occurs in 20–36% of patients.
- Gastrointestinal polyps and other abnormalities are present in about 72% of patients.
- Genitourinary tract involvement may include ovarian cysts and cancers.
- Central nervous system – development of Lhermitte–Duclos disease caused by hamartomatous growths of the cerebellum (rare).
- Skeletal abnormalities such as bone cysts.

What is the cause of Cowden disease?

Cowden disease is due to an abnormal PTEN tumour suppressor gene on chromosome 10q23. The PTEN protein product controls cell growth by promoting normal cell death. A mutation on the PTEN gene leads to loss of the protein's function and results in overproliferation of cells that form hamartomatous growths.

Cowden syndrome is an autosomal dominant condition meaning half of an affected person's children also have the syndrome.

How is the diagnosis made?

A diagnosis of Cowden disease can be made using the criteria below.

Major criteria	Minor criteria
<ul style="list-style-type: none"> • Breast cancer • Thyroid cancer, especially follicular thyroid carcinoma • Macrocephaly (very large head) • Lhermitte–Duclos disease 	<ul style="list-style-type: none"> • Other thyroid lesions • Mental retardation • GI hamartomas • Fibrocystic disease of the breast • Lipomas • Fibromas • Genitourinary tumours

Skin and mucous membrane lesions alone meet the criteria if

- Six or more facial papules are present, of which 3 or more must be trichilemmomas,
- Facial papules and oral mucosal papillomatosis are present,
- Oral mucosal papillomatosis and acral keratoses are present, or
- Six or more palmoplantar keratoses are present.

The diagnosis may also be made in the following circumstances:

- Two major criteria are met, but one must include either macrocephaly or Lhermitte–Duclos disease.
- One major and 3 minor criteria are met.
- Four minor criteria are met.

Diagnosis in a family in which one individual is diagnostic for CD is made when:

- Any single major criterion with or without minor criteria is met.
- Two minor criteria are met.

What is the treatment for Cowden disease?

Patients with Cowden disease need to undergo medical and physical examinations and appropriate laboratory and radiographic tests on a yearly basis to check for internal malignancies. Genetic counselling of relatives is very important especially females who are at most risk for malignant complications.

Treatment of the cutaneous features includes:

- Oral retinoids, e.g. [acitretin](#) that may temporarily control some of the cutaneous lesions. Lesions often reappear when treatment is stopped.
- Surgical care of facial papules using [chemical peels](#), [laser resurfacing](#), [surgery](#) and/or [shave excisions](#).

At least 40% of patients with Cowden disease have at least one cancer. If cancers are detected early their cure rate is high with appropriate treatment. Patients need to be followed-up regularly by a multidisciplinary team of doctors.

Related information

References:

- Book: Textbook of Dermatology. Ed Rook A, Wilkinson DS, Ebling FJB, Champion RH, Burton JL. Fourth edition. Blackwell Scientific Publications.

On DermNet NZ:

- [Cutaneous markers of internal malignancy](#)

Other websites:

- [Cowden Disease \(Multiple Hamartoma Syndrome\)](#) - emedicine dermatology, the online textbook

Books about skin diseases:

See the [DermNet NZ bookstore](#)

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DermNet does not provide an on-line consultation service.

If you have any concerns with your skin or its treatment, see a [dermatologist](#) for advice.

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