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Gardner syndrome

What is Gardner syndrome?

Gardner syndrome is a variant of the disease 'familial adenomatous polyposis' (FAP), an inherited disease that is characterised by gastrointestinal polyps, multiple osteomas (benign bone tumours), and skin and soft tissue tumours. Polyps tend to form at puberty with the average age of diagnosis around 25 years of age. In almost all patients, polyps will progress to malignancy, resulting in colorectal cancer so that timely detection is essential.

Gardner Syndrome is inherited as an autosomal dominant trait, so that an affected person has a 50% chance of passing on the gene to each of their children.

What are the clinical features of Gardner syndrome?

Clinical features of Gardner syndrome can be divided into two types, cutaneous and non-cutaneous. The most noticeable cutaneous feature of Gardner syndrome is the appearance of epidermoid [cysts](#). These cysts can be differentiated from ordinary epidermoid cysts by the following factors:

- Epidermoid cysts of Gardner syndrome occur at an earlier age (around puberty) than ordinary cysts
- Epidermoid cysts occur in less common locations such as the face, scalp and extremities compared to ordinary cysts
- Cysts tend to be multiple in over half of the patients with Gardner syndrome
- As with ordinary epidermoid cysts, cysts in Gardner syndrome are usually asymptomatic (without symptoms), however in some cases they may be pruritic (itchy) and/or inflamed, and they may rupture.

Other cutaneous features include fibromas, [lipomas](#), leiomyomas, [neurofibromas](#) and pigmented skin lesions.

Non-cutaneous features include:

- Gastrointestinal polyps that nearly always transform into colonic adenocarcinomas (colon cancer).
- Osteomas – these benign bone tumours are essential in making the diagnosis of Gardner syndrome. They occur most commonly in the mandible (jawbone) but may also grow in the skull and long bones.
- Dental abnormalities – as well as osteomas in the jaw there may be other dental abnormalities such as unerupted extra teeth and caries
- Multifocal pigmented lesions of the fundus in the eye – seen in 80% of patients. These lesions may be present shortly after birth and can be the first marker of the disease.

How is Gardner syndrome diagnosed?

Radiological studies are essential for patients and family members with suspected Gardner syndrome.

- Images of the long bone may show up osteomas.
- Images of the mandible at an early age may show up subtle defects.
- Eye exams at an early age can detect pigmented lesions of the fundus.
- Colonoscopy and other invasive tests to check for polyp involvement every 1–2 years.

What is the treatment for Gardner syndrome?

Treatment of epidermoid cysts in Gardner syndrome is similar to that used for ordinary cysts and involves [excision](#). Occasionally intralesional steroid injections may be used if the cysts are inflamed.

Surgical removal of gastrointestinal polyps is recommended because of the very high risk of polyps developing into cancer. Patients need regular colonoscopies.

Osteomas may also require excision only if they are severely deforming or if they are a nuisance.

Related information

On DermNet NZ:

- [Cysts](#)
- [Cutaneous markers of malignancy](#)

Other websites:

- [Gardner 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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