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LEOPARD syndrome

What is LEOPARD syndrome?

LEOPARD syndrome is a very rare inherited disorder that is characterised by skin, heart, ear, genital, head and facial abnormalities. The mnemonic LEOPARD describes these characteristic abnormalities associated with the disorder.

- L – lentigines (multiple brown–black spots on the skin)
- E – electrocardiographic (ECG) conduction defects
- O – ocular hypertelorism
- P – pulmonary stenosis
- A – abnormalities of genitals
- R – retarded growth resulting in short stature
- D – deafness or hearing loss due to inner ear malfunction

What are the clinical features of LEOPARD syndrome?

The clinical features of LEOPARD syndrome vary considerably between patients, with most only manifesting 3–5 abnormalities. [Lentigines](#) are the most common feature of the syndrome and occur in more than 90% of patients. However, they do not have to be present to diagnose LEOPARD syndrome.

The clinical features of lentigines are:

- Small, dark brown, round or oval, irregularly shaped flat spots 2–5 mm in diameter. However, some lentigines may grow to 1–1.5 cm in size.
- Often found on the face, neck, and upper part of the trunk but also on the palms, soles, and the sclerae (whites) of the eyes.

Careful examination of the skin usually shows up other skin abnormalities including:

- Freckling around the armpit region
- Café au lait spots (coffee–coloured flat birthmarks)
- Localised hypopigmentation (lightening of skin colour)
- Onychodystrophy (malformation of the nails)
- Interdigital webs (webbing between the fingers)
- Hyperelastic skin (stretchy skin)

Non–cutaneous features include:

- Hearing loss in about 25% of patients
- Short stature in about 1/3 of patients
- Mild degree of mental retardation in about 30% of patients
- Craniofacial abnormalities (e.g. low set ears, abnormal shaped skull, dental abnormalities)
- Abnormalities of the genitals in about 26% of patients, mainly in men
- Heart abnormalities, often without symptoms

What causes LEOPARD syndrome?

LEOPARD syndrome is nearly always due to mutations in the PTPN11 gene (protein-tyrosine phosphatase, nonreceptor type 11).

It is inherited in an autosomal dominant manner, which means that if one parent is affected there is a 50% chance that each child will be affected. About 70% of cases are inherited. The remainder are sporadic cases occurring from new mutations.

The signs and symptoms experienced by people with LEOPARD syndrome vary greatly. Some patients may have a partial form of the syndrome and suffer mild symptoms while others with the full syndrome are more severely affected.

How is LEOPARD syndrome diagnosed?

A proposed minimum criteria for the diagnosis of LEOPARD syndrome is:

- The presence of multiple lentigines, plus
- Features of at least 2 other categories
 - Other skin abnormalities
 - Cardiac abnormalities
 - Genitourinary abnormalities
 - Endocrine abnormalities
 - Neurologic defects
 - Craniofacial abnormalities
 - Shortness of stature
 - Skeletal abnormalities

If lentigines are absent, a diagnosis of LEOPARD syndrome may be made if the patient has at least 3 of the categories listed above and has an immediate relative also with the condition.

What treatments are available?

A team of specialist doctors including a cardiologist, endocrinologist, orthopaedist and dermatologist should manage LEOPARD syndrome.

If necessary, isolated lentigines can be removed through the use of [chemical peels](#), [cryotherapy](#), [laser treatments](#) or [surgical excision](#). For some patients, treatment with [topical retinoids](#) and [hydroquinone](#) cream may be helpful.

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:

- [Lentigo](#)
- [Brown spots and freckles](#)

Other websites:

- [LEOPARD syndrome](#) – e-medicine dermatology, the online textbook

Books about skin diseases:

See the [DermNet NZ bookstore](#)

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