REMARKABLY UNREMARKABLE

Morbus Fabry
Camouflaged by diversity

More information on **Fabry disease** is provided on the following websites:

- Information on Fabry disease, fabry-wissen.de
- International Network on Fabry, **fabrynetwork.org**
- Fabry centres Germany, **fabry-selbsthilfegruppe.de/fabry-zentren**

Please consult your doctor in case of any questions.

FABRY DISEASE FACTS

Cause
- Congenital accumulation storage disease
- Deficiency of a specific enzyme in the special substructures within each cell, the so-called lysosomes

Frequency of disease
- 1 in 40,000 men
- 1 in 20,000 women

Key symptoms
- Burning pain in hands and feet
- Little or no perspiration
- Reddish-purple skin rashes
- Gastrointestinal discomfort

- Impaired renal function
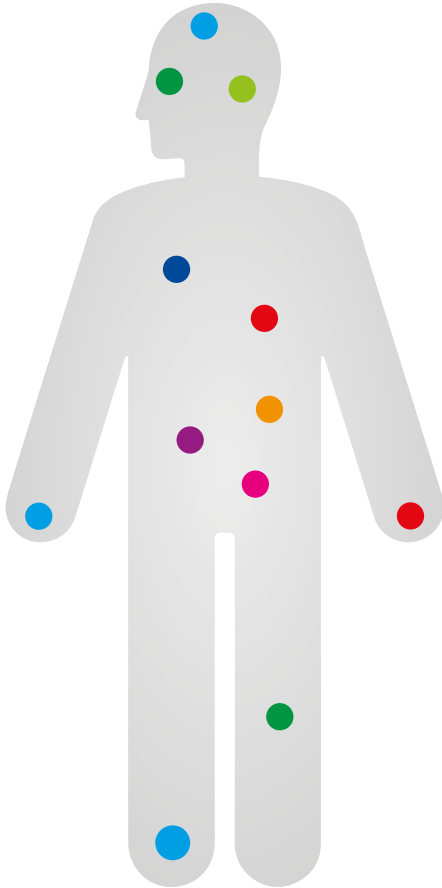- Cardiac arrhythmias
- Strokes in patients under 55

Diagnosis
- Men: Blood test
- Women: Gene analysis

Therapy options
- Enzyme replacement therapy (infusion)
- Chaperone therapy (capsules)

SYMPTOMS OF FABRY DISEASE

Brain
- Stroke

Mental discomfort
- Fatigue, listlessness
- Confusion
- Depression

Nervous system
- Burning pain on hands and feet
- Pain radiating into other parts of the body
- Joint pain
- Headache

Eyes
- Clouding of cornea/lens
- Alteration of vessels

Musculoskeletal system
- Uncoordinated gait
- Movement disorders

Ears
- Loss of hearing
- Tinnitus
- Dizziness

Lungs
- Shortness of breath
- Wheezing breathing sounds

Heart
- Shortness of breath due to enlarged heart
- Irregular heartbeat
- Palpitation

Nieren
- Frequent urination
- Proteinuria (in severe cases detectable by foaming)

Gastrointestinal tract
- Pain
- Diarrhoea
- Nausea, vomiting

Blutgefäße
- Fingers and sometimes toes turning white or blue

Skin
- Small, dark red skin elevations
- Oedema (swelling through water deposits)
- Dry skin, little or no perspiration
- Sensitivity to heat, fever