

# FABRY DISEASE SYMPTOM CHECKLIST








To be used in discussion with your doctor

Fabry disease is a rare, inherited disorder that can be passed from parents to children. It is caused by a mutation in the gene that controls an essential enzyme in the body's cells which can lead to progressive damage to the body including to the kidneys and heart.

## COULD YOU HAVE FABRY DISEASE?

Each Fabry disease patient can be affected in a different way, and can therefore experience varying degrees and combinations of symptoms. These symptoms arise over time as a result of the gradual build-up of a fatty substance known as Gb<sub>3</sub> in the cells and tissues. This checklist is a compilation of some of the typical signs or symptoms of Fabry disease that you can print out and take to your doctor if you are experiencing some, or a combination of these symptoms. The list is not intended to be exhaustive.

**Please complete the checklist and provide as much detail as you can to help your doctor.**

	Symptoms (more than one can be selected)		Family history of this symptom (please indicate relative affected)
<b>EYE</b>	<b>Yes</b>	<b>No</b>	
 Clouding of the cornea of your eye, without it affecting your vision	<input type="radio"/>	<input type="radio"/>	<input type="radio"/> .....
<b>EAR</b>	<b>Yes</b>	<b>No</b>	
 Hearing loss	<input type="radio"/>	<input type="radio"/>	<input type="radio"/> .....
Ringing in ears	<input type="radio"/>	<input type="radio"/>	<input type="radio"/> .....
<b>SKIN</b>	<b>Yes</b>	<b>No</b>	
 Small skin marks of red to blue colour between the belly button and knees (angiokeratoma)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/> .....
Decreased sweating	<input type="radio"/>	<input type="radio"/>	<input type="radio"/> .....
Heat or cold intolerance	<input type="radio"/>	<input type="radio"/>	<input type="radio"/> .....
<b>NEUROPATHIC PAIN</b>	<b>Yes</b>	<b>No</b>	
 Burning pain in hands	<input type="radio"/>	<input type="radio"/>	<input type="radio"/> .....
Burning pain in feet	<input type="radio"/>	<input type="radio"/>	<input type="radio"/> .....
<b>FATIGUE</b>	<b>Yes</b>	<b>No</b>	
 Tiredness or fatigue (which can be extreme)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/> .....
<b>GASTROINTESTINAL</b>	<b>Yes</b>	<b>No</b>	
 Abdominal cramping	<input type="radio"/>	<input type="radio"/>	<input type="radio"/> .....
Frequent bowel movements shortly after eating	<input type="radio"/>	<input type="radio"/>	<input type="radio"/> .....
Diarrhoea	<input type="radio"/>	<input type="radio"/>	<input type="radio"/> .....
Nausea	<input type="radio"/>	<input type="radio"/>	<input type="radio"/> .....
<b>GENERAL</b>	<b>Yes</b>	<b>No</b>	
 Depression	<input type="radio"/>	<input type="radio"/>	<input type="radio"/> .....

In addition to the common signs and symptoms seen overleaf, there are more serious symptoms which may indicate the presence of Fabry disease. It is important to manage the disease appropriately to help minimize long-term complications. You may have been told that you have some of these conditions by a doctor.

**Please complete the checklist and provide as much detail as you can to help your doctor.**

**Symptoms**  
(more than one can be selected)

**Family history of this symptom**  
(please indicate relative affected)



**KIDNEY**

**Yes**      **No**

- Unexplained kidney problems                   .....
- Proteinuria (excess protein in the urine)                   .....
- Dialysis                   .....



**HEART**

**Yes**      **No**

- Unexplained heart problems including changes in the shape and function (enlarged left ventricle)                   .....
- Angina                   .....
- Arrhythmia or irregular heart beat                   .....
- Heart attack                   .....
- Exercise intolerance                   .....



**BRAIN**

**Yes**      **No**

- Stroke                   .....
- Dizziness                   .....

If you are suffering from any other symptoms that are not listed above and that you would like to discuss with your doctor, **please make a note of these below:**

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If a combination of the above applies to you, or have been told previously by a doctor that you have any of these conditions, it is possible that you may have Fabry disease. Please note that these symptoms and combination of symptoms do not qualify as a diagnosis of Fabry disease. Please refer to your doctor for further advice.

**Ask your doctor for further information**

**Visit [www.fabrydisease.info](http://www.fabrydisease.info) for more information**