

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. This checklist is a compilation of some of the typical 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.

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)
		Yes	No	
	Ear			
	Hearing loss	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Ringing in ears	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Skin	Yes	No	
	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"/>
	Neuropathic pain	Yes	No	
	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"/>
	Fatigue	Yes	No	
	Tiredness or fatigue (which can be extreme)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Gastrointestinal	Yes	No	
	Abdominal cramping (often after eating)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Diarrhoea	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	Nausea and vomiting	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
	General	Yes	No	
	Depression	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

In addition to the common symptoms seen overleaf, there are more serious symptoms which may indicate the presence of Fabry disease. These symptoms arise over time as a result of the gradual build-up of fatty substances called glycosphingolipids in the cells and tissues of the body.

If left untreated, vital organs eventually start to deteriorate and severe conditions can arise. You may have been told that you have some of these conditions by another doctor.

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

	Signs and symptoms (more than one can be selected)		Family history of this sign/symptom (please indicate relative affected)
	Yes	No	
Kidney			
Unexplained kidney problems	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Excess protein in the urine (proteinuria)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Dialysis	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Heart			
Unexplained heart problems including changes in the shape and function (enlarged left ventricle)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Angina	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Arrhythmia or irregular heart beat	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Heart attack	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Exercise intolerance	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Brain			
Stroke	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Dizziness	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

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:

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 for more information

