

The Boston Refsum Center of Excellence at MGH provides care for children, adolescents and adults with Refsum Disease or Refsum-like disorders. Adult Refsum disease is a metabolic disorder caused by a number of faulty genes (principally PHYH) which affect the pathway that degrades a plant-derived chemical called phytol which some bacteria found in ruminant animals and fish turn into Phytanic Acid. In humans if phytanic acid is not degraded it accumulates in nerves, liver and fat. Phytanic acid is not produced in the body but found in some foods as a result of the bacterial fermentation of green plants or algae. The highest concentration of phytanic acid is found in dairy products, beef, lamb and some fish products. Refsum disease can be life threatening if undiagnosed.

We integrate state-of-the-art resources with compassionate, comprehensive care. National experts from Massachusetts General Hospital and Massachusetts Eye and Ear Infirmary work together to provide care for people with Refsum Disease.

[See Dr. Florian Eichler's comments on Refsum Disease.](#)

Our multidisciplinary approach includes the following specialists:

- neurologist
- neuromuscular specialist
- ophthalmologist
- dietician
- Otolaryngologist (ENT)
- audiologist
- metabolic specialist
- psychiatrist
- genetic counselor

We also work closely with all of the other departments and can make specific referrals, as needed. For your convenience, a central appointment coordinator is available to schedule your appointments. Please contact Natalie Grant at ngrant@mgh.harvard.edu

For more information on Refsum Disease, go to:
<https://www.defeatadultrefsumeverywhere.org/>

