



**Global DARE Foundation** is a registered non-profit public charity.

Our mission is to promote world-wide awareness and better quality of life for all who are diagnosed with Adult Refsum Disease.



**HELP US GET  
REFSUM PATIENTS  
DIAGNOSED  
EARLIER, IMPROVE  
QUALITY OF LIFE  
AND FIND A CURE!**



## **CONTACT / DONATE**

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***We DARE to believe in a  
cure for Refsum Disease***



## **REFSUM DISEASE**

Adult Refsum disease is a genetic disorder that impacts the metabolism of phytanic acid.

It is one of the few causes of Retinitis Pigmentosa that has a **proven dietary therapy to slow vision and hearing loss.**



**DefeatAdult  
RefsumEverywhere.org**

# CLINICAL FEATURES OF REFSUM DISEASE

## CURRENT THERAPIES

### Low Phytanic Acid Diet

A strict diet low in phytanic acid is used for long-term management. This diet includes the exclusion of foods that are high in phytanic acid such as dairy products, beef, lamb and some fish.

### Plasmapheresis

A dialysis-type process that is the direct removal of phytanic acid in the blood. Typically used in acute management and occasionally used for long term management.

### Symptom Management

Symptom management could include cataract surgery, eye drops, hearing aids, cochlear implants, skin lotions, pain management, knee replacement, heart and liver transplant.



Retinitis Pigmentosa  
(loss of vision)



Loss of Smell



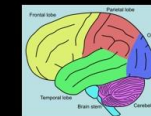
Peripheral Neuropathy  
(numbness, nerve pain)



Loss of Hearing



Bone Abnormalities



Cerebellar Ataxia  
(balance issues)



Ichthyosis  
(itchy, scaly skin)

**Other Symptoms include:** cataracts, nystagmus, small pupils, skeletal dysplasia, cardiac arrhythmia, weakness



## PATIENT REGISTRY

A patient registry will bring the Refsum community and researchers together to gain a better understanding of Refsum Disease and drive better therapies.

**ENROLL TODAY** at the [Global DARE Foundation website](#).



## DIAGNOSIS

Diagnosis of Refsum Disease can be done through the following:

- Clinical exam
- Phytanic Acid Level
- Fibroblast Test
- Genetic Testing

Additional information and videos are available at the [Global DARE Foundation website](#)