



Relationship Between Hearing Loss and Adult Refsum's Disease

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Introduction

Adult Refsum's disease is a rare autosomal recessive disorder inherited from both parents. Individuals who are effected by this genetic condition are unable to break down phytanic acid (PA) in the blood. This is a fatty acid found in food such as dairy, animal fats and some fish.

This inability to break down phytanic acid causes the acid to build up in the tissues and blood leading to side effects such as retinitis pigmentosa (blindness), hearing loss, anosmia, skin changes and vestibular disorders.

Question

What is the relationship between adult Refsum's disease and hearing loss, and can it be managed by changes in diet?

Search Terms

PICO Question

Search Terms

P Adult Refsum's disease

Search Terms Adult, Adults, Refsum, Refsum's, disease, disorder

I Dietary management

Search Terms Dietary, diet, intake, management, food,

C Lack of treatment via dietary intake

Search Terms Lack of management, dietary neglect

O Minimization of effects of hearing loss

Search Terms Hearing, hearing loss, sensorineural hearing, ear, loss

Inclusion / Exclusion Criteria

The following criterion were chosen to systematically narrow the search results based on my research question

Inclusion Criterion:

Exclusion Criterion:

- Subjects are human
- Subjects are adult (18 years+)
- Subjects have Refsum's Disease (adult)
- Hearing loss is discussed as a side effect of Refsum's disease
- Discusses dietary management
- Accessed through UWO Libraries

- Non-human subjects
- Infantile Refsum's disease
- Children as subjects
- Not available in English
- Not available via Western Libraries
- Duplicates
- Some articles were excluded based on relevance after reading the abstract

Literature Search

A systematic review of the literature was conducted. Search terms were run through many databases including PubMed, CINAHL, Scopus and Ovid. A secondary search was done based on the references of the used in the relevant studies.

5
After reading and data extraction

Results

Five relevant articles were chosen to critically review using the Crowe Critical Appraisal Tool (CCAT). A data extraction tool was used to highlight the key findings and information provided in each study. Research design, research methods, subject characteristics, sample size, intervention implemented, outcomes found, statistical significance, conclusions and the CCAT quality score were used to extract the data and evaluate the current literature available. The data extracted from the five articles has been summarized and is displayed on the table below.

#	Research Question/Objective	Sample Size	Intervention Description	Results	Conclusions	Quality Score
1	What is the site of origin of hearing loss in those with adult Refsum's Disease?	9	Total audiologic examination. ABR, tympanometry, pure tone audiometry, acoustic reflexes on those with diagnosed adult Refsum's disease based on serum phytanic acid blood levels.	Mild-moderate mostly high frequency hearing loss in 7/9 subjects. Found subtle auditory nerve involvement in 6 of 7 subjects with hearing loss and 1/ of 2 normal hearing individuals.	Patients with adult Refsum's disease often report hearing difficulties. Auditory Neuropathy is also common in this cohort. They should have complete audiometric investigations including tympanometry, pure tone audiometry, ABR and OAE's so that appropriate audiologic rehabilitation is provided.	77% (31/40)
2	To describe a case of bilateral cochlear implantation on a 59 year old individual with vision, hearing loss, and tinnitus due to adult Refsum's disease	1	The individual's pure tone audiogram has deteriorated over a 10 year period to having no response to the limits of the audiometer or ABR. Testing post implantation included pure tone audiometry, BKB-SIN, SRT, and evaluation of tinnitus symptoms.	After cochlear implantation, the patient demonstrated improved audiometric performance and improved sound localization. SRT was 25 dB and BKB scores were 98%. Two years after unilateral implantation, the individual was implanted on the other ear. The patient reported increased localization abilities, hearing in noise and speech discrimination. Subjective tinnitus also improved with the second implant.	Bilateral cochlear implantation is an important strategy in the improvement of hearing and quality of life for individual's with adult Refsum's disease. Especially when vision loss is a common co-morbid disorder that accompanies Refsum's disease, there is an increased reliance on the auditory system for spatial awareness. Cochlear implantation helped this patient with hearing as well as subjective tinnitus. This case study provides information and support for CI's in adult Refsum's disease.	57.5% (23/40)
3	Report on experience of two patients with adult Refsum's disease who underwent cochlear implantation. Also aims to highlight the potential anesthetic issues that a patient with Refsum's disease poses.	2	Report on each patient's experience with cochlear implantation including SRT's, BKB SIN, BKB in quiet and quality of life questionnaires over time.	One patient was able to hear, identify environmental sounds, use the telephone and engage in conversation with minimal background noise at three months. By nine months "nearly everything could be heard". She was especially dependent on hear cochlear implant and auditory stimulation since by two years her eyesight deteriorated. The other patient received bilateral cochlear implants and by three months was able to communicate successfully over telephone and had impressive abilities to identify words in noise	Cochlear implants are useful to consider in Refsum's disease since these patients are often deprived of other senses such as sight and smell. Especially if an individual is visually impaired in addition to profound hearing loss (dual sensory loss). Bilateral cochlear implantation has the potential to give the individual back localization of sound, hearing in quiet, and speech in noise. These benefits can not only increase communication for these individuals but is important to their personal safety.	67.5% (27/40)
4	Objective is to identify the site of hearing loss in Refsum's disease	1	DPOAE's, ABR, pure tone audiometry, monitoring dietary intake over the span of about 10 years.	The individual had poor compliance with dietary restrictions and his hearing loss progressed from near normal hearing and robust OAE's to mild loss and poor ABR results and finally progressing to profound hearing loss, no ABR results however robust OAE's. This indicated the presence of auditory neuropathy in this individual	This study is the first case of Refsum's disease to examine both ABR's and OAE's along with standard pure tone audiometry. The authors conclude the likely site of lesion in this case is post-outer hair cell and warn about the dangers of amplification for those with auditory neuropathy. The authors also note the importance of dietary management in the amount of phytanic acid consumed as it may prolong presentation of hearing loss in those with diagnosed Refsum's disease.	75% (30/40)
5	What is the long term efficacy in reducing symptoms of Refsum's disease	13	A retrospective audit was conducted for subjects who had attended the Refsum's clinic for 10 years or more. Information on body weight, PA level, symptoms of adult Refsum's disorder and illness from case notes. PA was also measured. Data was analyzed by constructing time series plots of PA for each patient.	Dietary therapy reduces PA levels by 89%. 30% of the patients were able to manage their PA levels. None of the patients developed acute complications requiring plasmapheresis during the study and significant PA levels decreased.	Regular dietary review and education on dietary restriction for those with PA accumulation can be effective at managing and overall reducing the PA for many years. Regular review may be key to achieving and managing a sustained reduction in PA levels..	87.5 (35/40)

Conclusions

Data extracted from the five research studies above demonstrates that hearing loss commonly presents in individuals with adult Refsum's disease. There are several presentations of hearing loss in these individuals ranging from progressive sensorineural hearing loss to auditory neuropathy spectrum disorder. Literature on Refsum's disease and site of lesion for hearing loss is very limited. Individuals with adult Refsum's disease should certainly have a complete audiological assessment to determine the nature of the hearing loss and appropriate form of rehabilitation, whether it be amplification, counseling or cochlear implantation. Vision, and olfactory degeneration is also commonly seen in those with adult Refsum's disease, making auditory rehabilitation critical to giving these individuals one of their main senses. Hearing loss due to Refsum's disease can sometimes be prolonged to a later onset by strict dietary management avoiding dairy and animal meat, but there is not a cure for adult Refsum's disease nor the hearing loss than accompanies it. More research and education is needed for individuals with adult's Refsum's disease for early intervention in their diet and hearing health.

References

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