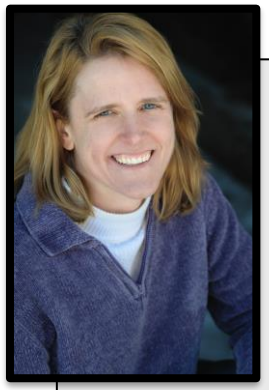
REFSUM DIET OVERVIEW

Global DARE Foundation

#CureRefsum

Today's Hosts & Presenters

Hosts



Kristie DeMarco

President and Founder at Global DARE Foundation

Susan Kuranoff

Secretary and Co-Founder at Global DARE Foundation

Presenters

Eleanor Baldwin

Clinical & Research Dietitian at Guy's & St. Thomas Hospital

Sarah Firman

Specialist Dietitian at Guy's & St. Thomas Hospital

Today’s Agenda

- Global DARE Foundation’s Mission
- Refsum Diet Overview
- Question & Answer Session

Webinar Housekeeping Details

- All participants are in listen only mode
- How to ask a question during the Q&A:
  - Participants following on Zoom can type their questions in the Q&A box at any time during the presentation or by raising their hand at the end to ask a question live.
  - Participants joining by phone can press *9 on their phone to raise their hand.
- Questions will be answered in the following order:
  - Q&A box in Zoom
  - Dial in participants
  - Online participants
- Today's session will be recorded for later viewing on Global DARE Foundation Website (www.defeatadultrefsumeverywhere.com)

Global DARE Foundation
#CureRefsum

DEFEAT
ADULT
REFSUM
EVERYWHERE

DARE'S MISSION

Global DARE Foundation's mission is to promote world-wide awareness and better quality of life for all who are diagnosed with Adult Refsum Disease.

Nutrition – A tool to manage Adult Refsum's Disease

Eleanor Baldwin & Sarah Firman

Specialist Dietitians in Adult Refsums Disease

Aims of Our Presentation

Provide practical dietary advice that people with ARD and their families can use in their daily lives.

Improve the knowledge of health care professionals working with people with ARD.

Generate an awareness of gaps in research and information so that future research can be targeted towards improving the quality of life of people with ARD.

What is the evidence on which we base our dietary advice?

- Research on the biochemistry behind ARD.
- Research on the composition of food and how that changes in response to alterations in growing conditions, animal feeding and slaughtering practices, the development of new foods and recipes and changes in food labelling guidelines.
- Knowledge of human physiology and normal metabolism during rest, fasting and different types of exercise.
- Information and knowledge gained from the experience of people with ARD and the dietitians who work with them in the UK and throughout the world.

What Are The Lessons Learnt From Biochemical Research?

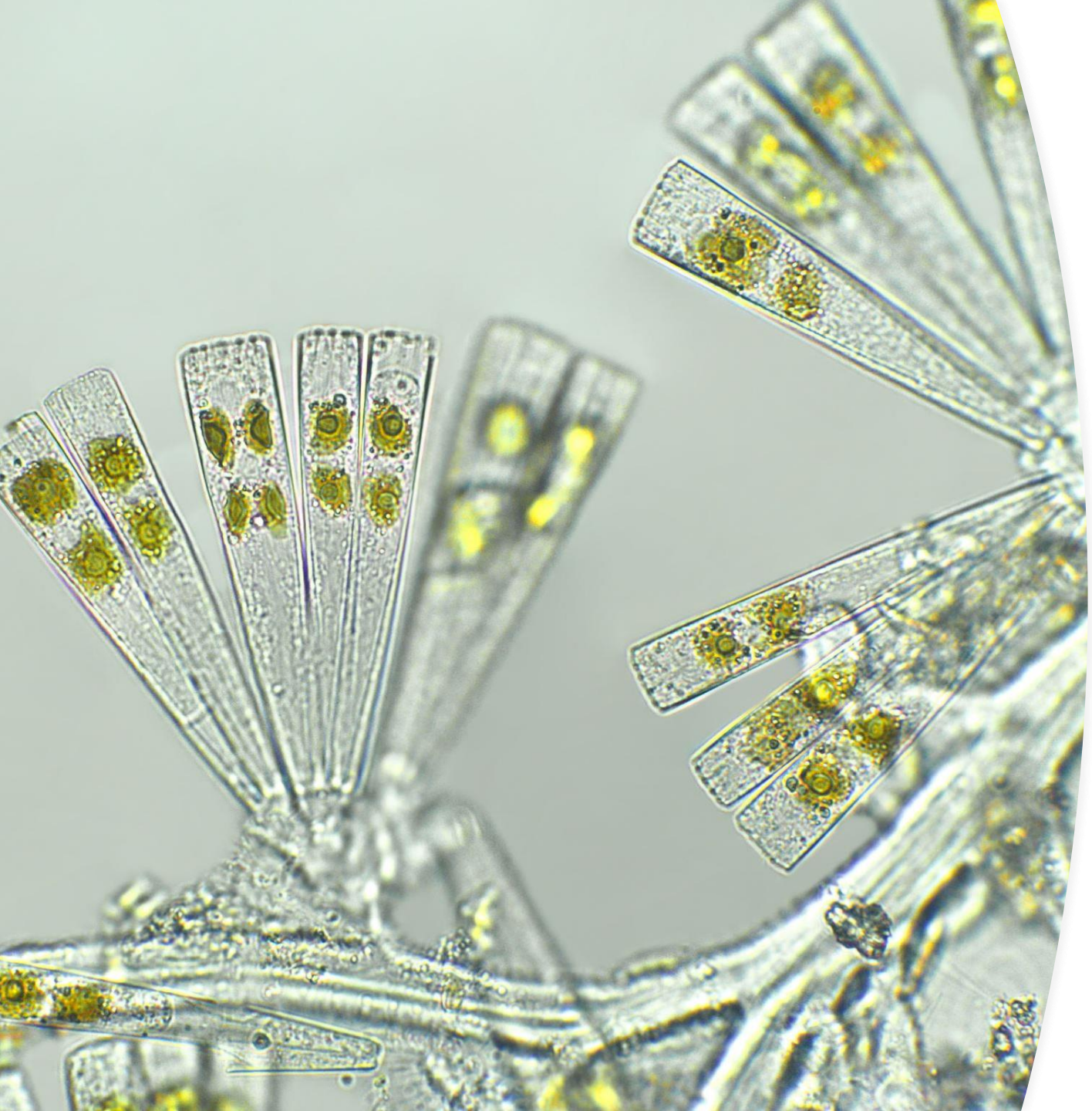
1. People with ARD have a reduced ability to metabolise/breakdown one fatty acid called phytanic acid.

   This fat only comes from the diet
   The main breakdown pathway (alpha oxidation) does not function
   A minor pathway, called omega oxidation still works

What Does This Tell Us?

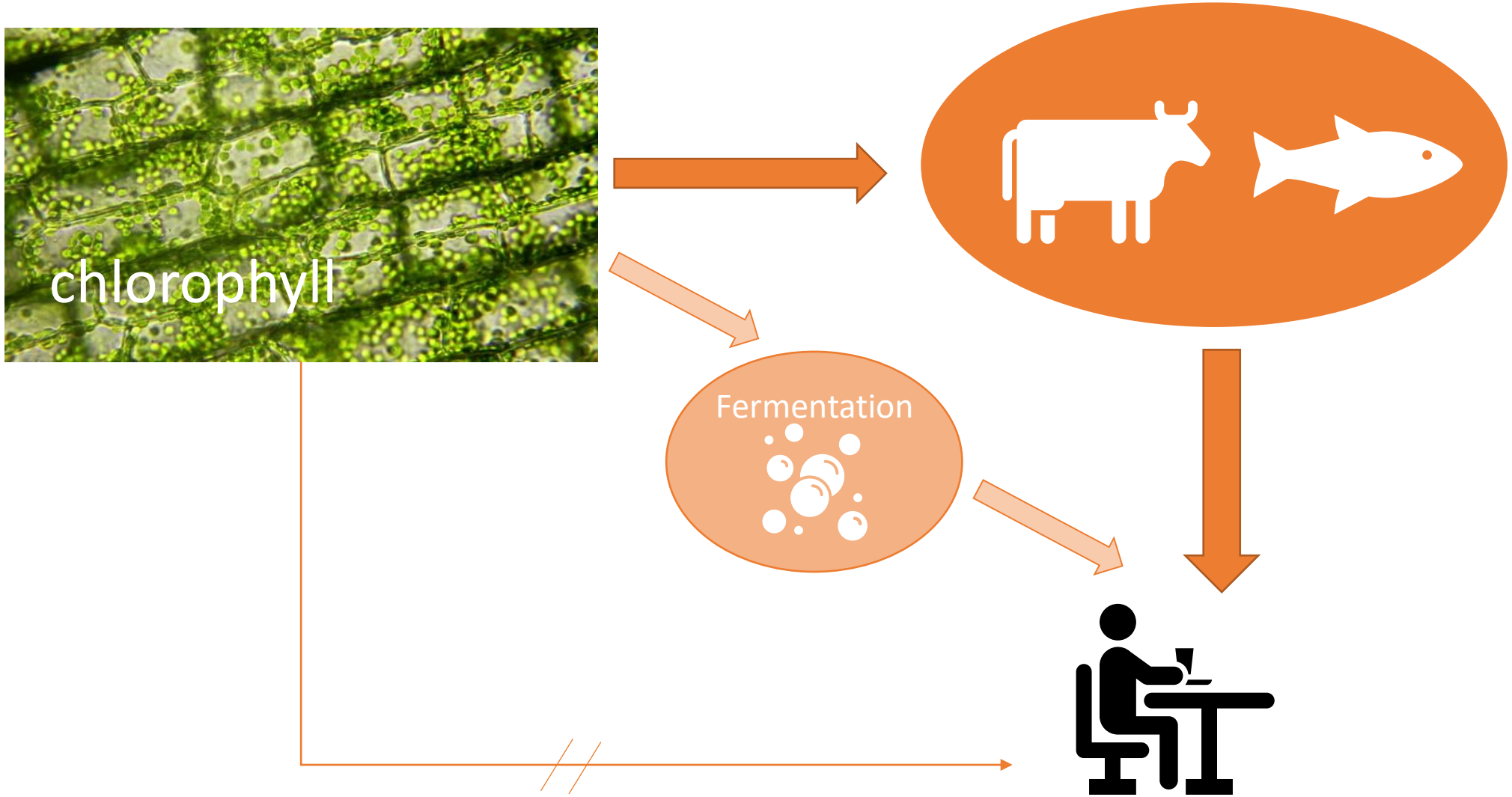
WE NEED TO KNOW WHERE PHYTANIC ACID COMES FROM IN THE DIET SO THAT WE CAN AVOID RICH SOURCES

WE NEED TO UNDERSTAND IF OTHER SUBSTANCES IN FOOD CAN BE CONVERTED INTO PHYTANIC ACID



Where Does Phytanic Acid Come From?

- Phytanic acid is a breakdown product of chlorophyll.
- It is produced in large amounts by ruminant animals. This means that cows, sheep, goats and their meat and dairy products are rich sources.
- Fish contain phytanic acid which comes from algae or eating other fish which have eaten algae.
- Fermented vegetable products such as kimchi and sauerkraut are presumed to contain phytanic acid.


chlorophyll
Fermentation

More Biochemistry..

Omega oxidation produces a substance called 3 methyl adipic acid. The production of this compound has been measured and this led to the conclusion that people with ARD are able to process some phytanic acid and that their blood concentration of phytanic acid will gradually reduce if intake is **limited to 10mg of phytanic acid** a day.

Which Food Substances are Converted to Phytanic Acid in the Body?

Free phytol can be converted to phytanic acid. Free phytol is found in small amounts in many fruit and vegetables. Previous studies suggested that free phytol contributes less than 10% of the phytanic acid in the blood. Further research is required.

Phytyl fatty acid esters have recently been found to be converted to phytanic acid in the body. Phytyl fatty acid esters have been found in: red and yellow bell peppers, rocket, green olives, red grapes, carrots and cucumber.

Reference: Krauß S, Michaelis L, Vetter W (2017), PLOS ONE 12(11)

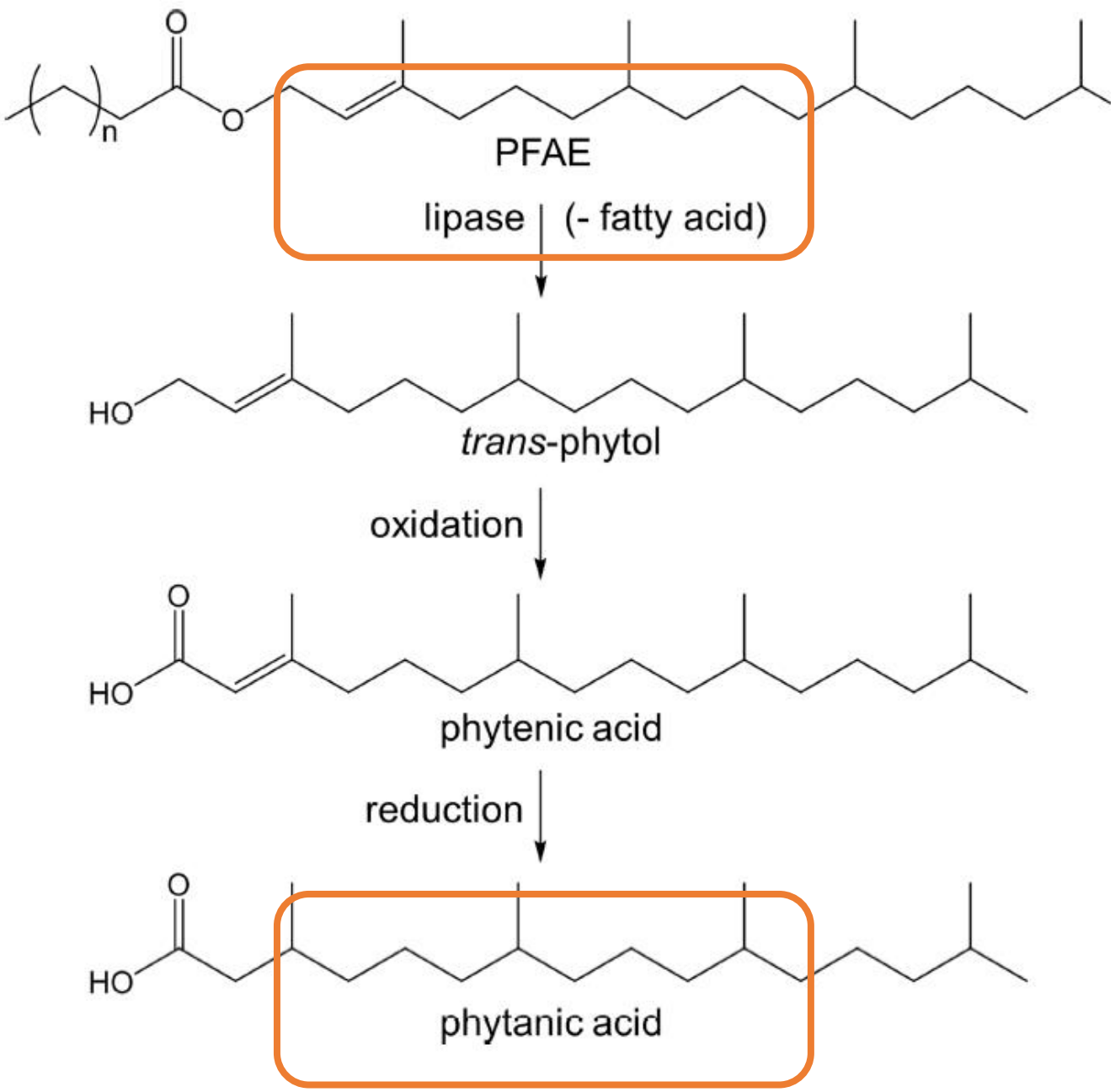


Image from: Krauß S et al (2017)

Can I still eat these foods?

Phytyl fatty acid esters

1 handful of rocket

Recommend to avoid
and have green peppers

These foods contain only very small
amounts and do not need to be restricted

Black olives do not
need to be limited

More research needed

Analysis of phytanic acid content in a range of foods

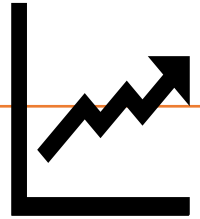
Analysis of a broader range of fruits and vegetables

Understanding the role of different portion sizes and food groups on overall phytanic acid intake

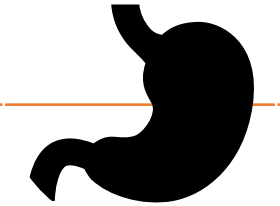
2. Phytanic acid that is not broken down is stored in the liver, adipose tissue and all other tissues and organs containing fat

What Does This Tell Us?

Circumstances that cause the release of phytanic acid from stores will lead to a rapid rise in phytanic acid in the blood.



Fasting: Phytanic acid doubling time in fasting is 29 hours.



Rapid weight loss caused by surgery, poor intake, excessive exercise or emotional distress.

Illness with fever, vomiting, diarrhoea or loss of appetite and intake.

This means..

THE PHYTANIC ACID CONCENTRATION IN THE BLOOD WILL GRADUALLY FALL IF THE INTAKE OF PHYTANIC ACID AND THE FOOD SUBSTANCES THAT ARE CONVERTED TO PHYTANIC ACID IN THE BODY ARE KEPT BELOW THE ACTIVITY OF THE OMEGA OXIDATION PATHWAY

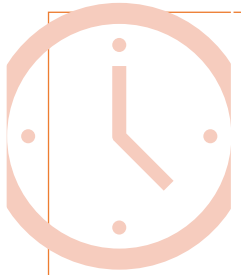
IF

LARGE AMOUNTS OF PHYTANIC ACID ARE NOT RELEASED FROM BODY STORES

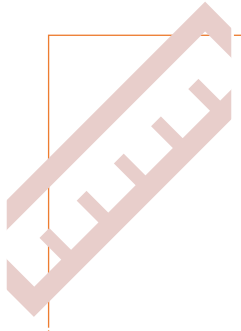
Effect of metabolism on ARD

- Glucose is the preferred fuel for many parts of the body, including the brain.
- In order to maintain the blood glucose within its desired range the body will create glucose from its liver and muscle glycogen stores during fasting and intense or prolonged aerobic exercise.
- Prolonged fasting, following a low carbohydrate diet and prolonged anaerobic exercise increase fat oxidation and the **release of stored phytanic acid from the liver**

How do I limit the release of phytanic acid from body stores?



Regular meals are important. Extended fasting is not recommended.



Avoid rapid weight loss. Weight loss if you have a high serum phytanic acid is not recommended.

Ensuring adequate glucose is important during times of illness.

Practical Tips

Always eat breakfast.

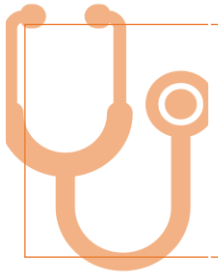
Eat carbohydrate before exercise (banana, biscuit, fruit, cereal bar etc).

Eat regular meals and avoid large gaps between meals.

Eat carbohydrate at each meal, preferably wholegrain or higher fibre varieties.

Metabolism is Faster in Physical and Mental Illness

- Illness causes the production of stress hormones which are catabolic
- This means that glycogen and fat stores are released more quickly
- In addition, illness can lead to poor appetite and intake



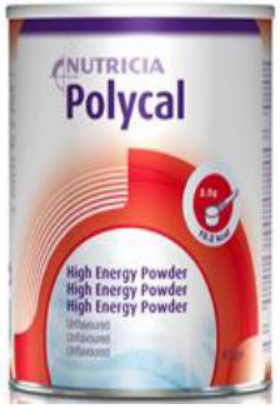
Ensuring adequate glucose is important during times of illness

Illness can be an Emergency

- A regular source of glucose through nutritional supplement drinks or an emergency regimen should be started immediately when unwell
- Contact your medical team
- If you are admitted to hospital for planned or emergency treatment, provide your health care professionals with your emergency regimen details and contact information for your treating physician

Our Emergency Regimen

- Glucose drink (20% glucose concentration) during illness



Our Emergency Regimen

Stage 1	Stage 2	Stage 3
At first sign of feeling unwell/loss of appetite, take 200ml of 20% glucose drink If better within 1 hour return to a usual diet If unwell, follow stage 2 or 3	Commence full emergency regimen, taking 200ml glucose drink every 2 hours Continue to eat as tolerated Contact your metabolic service	If unable to tolerate emergency regimen, if not improving after 24-48 hours, or if you become increasingly unwell, present to hospital and inform your metabolic team. IV glucose will be required

Putting the science into the everyday…

Food swaps

Beef, lamb, sheep and goat

Choose chicken, turkey, pork, ham or soya substitutes and tofu instead

Oily fish such as salmon and tuna

Choose cod, coley, haddock, crab and prawns instead

Changes in Food Composition

The amount of phytanic acid in ruminant animal products and fish is affected by:

- What the animal or fish is fed.
- Organic or not organic.
- The cut of the meat chosen and the carcass grading criteria used.
- The fat content of the product.
- How restaurant dishes are “finished”.
- Changes in ingredients used in mixed dishes.

Practical Tips

- Organic versions of ruminant animal products contain much more phytanic acid than non-organic versions.
- Cuts of meat and dairy products with a high fat content will have the highest phytanic acid content.
- Check ingredient labels for change regularly.
- Check if restaurants “finish” vegetables, pasta, potatoes, fish or rice with butter.
- Fat free does not mean absolutely no phytanic acid (fat free yoghurt is high phytanic acid).

Food swaps

Full fat, whole milk, semi-skimmed milk

Skimmed or skim (0.1% fat) milk

Cheese (cows, goat or sheep)

Vegan cheese – soya, coconut cheese

Fat free quark and fat free fromage frais

Food swaps

Butter

Vegetable oil based spreads

Yoghurt (cows, goat, sheep)

Soya, oat or coconut yoghurt

Fat free yoghurts should be avoided

Changes in Eating Behaviour

- Increasing emphasis on plant based eating and sustainable food.
- Increase in the number of meals eaten outside the home.
- Increase in new food products – plant milks, vegan cheeses, vegan meats.
- Diversifying food shopping habits – recipe boxes, foods from farmers markets, home grown food, the globalisation/localisation of food production, less /more cooking from basics.
- This may all change post COVID-19.

Practical Tips

- Vegan products should be safe choices – but food testing is needed for phytanic acid, Phytol and Phytyl fatty acid esters.
- Where vegan products are used it is important to ensure that a nutrient shortfall does not occur (e.g. nut milks and calcium and protein status).
- Vegan products are often lower in energy – avoid weight loss unless your serum phytanic acid is low (less than 200umol/l).

Low in phytanic acid

Vegetables and salad
(portion sizes for those containing free phytol and phytyl fatty acid esters)

Fruits

Low in phytanic acid

Rice, pasta, bread and breakfast cereals (check ingredients for milk powder)

Practical Tips

- Essential fatty acids: **Omega 3 and 6**
  - Found in vegetable oils such as walnut, flaxseed (linseed), sunflower, soybean and canola oils
  - **Omega 6**: poultry, eggs, wholegrain breads

  - Omega 3 is converted to eicosapentaenoic acid (EPA), and docosahexaenoic acid (DHA)
  - DHA and EPA have the most direct health benefits
  - The main source of EPA and DHA are marine oil/oily fish, which are high in phytanic acid, and need to be avoided.

Practical Tips

- Discuss with your dietitian about reviewing your omega 3 and 6 intake to ensure it meets your needs
- For those with increased requirements (pregnancy and lactation) additional DHA/EPA supplementation may be required

- Most over the counter omega 3/EPA/DHA supplements have **not** been tested for phytanic acid content or contain fish oils.
- Vegan options can be made with algae and may be high in phytanic acid

Practical Tips

- Omacor has been tested for use on low phytanic acid diet
- KeyΩmega™ or docΩmega™ are vegetable oil based and could be considered on a low phytanic acid diet.

- More research is needed into the phytanic acid content of essential fatty acid supplements.

What Have We Learnt From People with ARD?

- There is no such thing as a “normal portion size”.
- Vitamin & mineral deficiencies can occur.
- People change their dietary habits with sometimes unforeseen results.
- Lack of a sense of smell creates a dependence on the four main tastes for flavour. This can result in a lack of interest in food and a high salt intake.
- People vary in their response to diet.

Top Tips

- Focus on temperature, sweet, sour, texture and spice to enhance the flavour and enjoyment of food.
- Have your vitamin D status checked annually. Vitamin B12, folate and iron status may also need checking.
- Discuss with your dietitian about your intake of essential fatty acids
- Discuss a desire to lose weight or make major changes to your diet in case the consequences are likely to be poor.
- Contact your medical team prior to surgery and during illness.

Global DARE Foundation
#CureRefsum

Q&A

For more information contact:

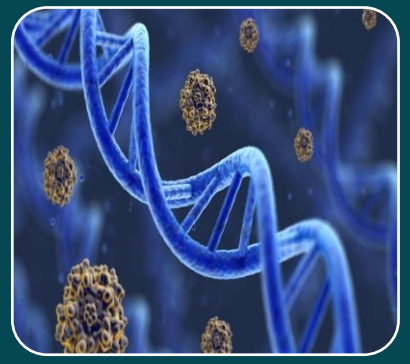
Global DARE Foundation

info@globaldarefoundation.org

www.defeatadultrefsumeverywhere.org

UPCOMING REFSUM WEBINARS

Global DARE Foundation will be holding additional webinars throughout the summer. Registration can be accessed through our website at https://www.defeatadultrefsumeverywhere.org/dare-events



8/7/20, 8:00 PM EST

Gene Therapy - A Potential Therapy for Refsum Disease

Ryan Butler, PhD from UT Southwestern will provide an overview of Gene Therapy as a potential future therapy for Refsum Disease