Released January 2024

Dietary information for the management of Adult Refsum disease

What is Adult Refsum disease?

Adult Refsum disease is a rare condition affecting approximately one in a million people. People are born with this condition because they inherited a faulty gene from both of their parents. The gene affects the body's ability to process a type of fat found in the diet called phytanic acid. Phytanic acid builds up in the body from eating foods high in phytanic acid, or from situations that cause the body to break down fat stores, releasing phytanic acid, including fasting, illness, low carbohydrate diets, exercise without appropriate fuel, weight loss and high caffeine intake

The levels of phytanic acid build up in the body over time and eventually cause symptoms of worsening vision, hearing and sense of smell. There can be a loss of sensation in the feet and legs. There may be difficulty in coordinating walking and balance. The skin can become itchy and scaly as well as other symptoms.

What is the treatment for Adult Refsum disease?

Treatment aims to reduce the phytanic acid content of the blood through diet and, if necessary, by a treatment called plasmapheresis. When the phytanic acid content in the blood falls, there can be a return of sensation to the feet and legs, improvements in walking and balance and the skin can return to a healthy state. Damage to the eyes, hearing and sense of smell cannot currently be reversed, but further loss is generally slow to occur.

How does the Westminster Refsum's Diet work?

The Westminster Refsum's Diet has been shown to be effective at lowering the amount of phytanic acid in the blood. The diet works by:

1. Restricting foods high in phytanic acid.

2. Reducing the release of phytanic acid from body stores with having regular meals containing carbohydrates, getting enough energy (calories), fuelling appropriate around exercise, managing illness with an emergency regimen, preventing unplanned weight loss and limiting caffeine.

How quickly will my phytanic acid fall?

The phytanic acid content of the blood will gradually decrease over time. The decrease is not normally a straight line and the rate of decline varies from person to person.

Where does phytanic acid come from?

Phytanic acid is present in larger amounts when phytol, which is part of the chlorophyll molecule, is fermented by bacteria. This occurs in the stomach of ruminant animals (cows, sheep).

There are also two substances that can be converted to phytanic acid by the body: phytyl fatty acid esters and free phytol. Both of these substances come from plant-based foods such as vegetables, fruit, nuts, seeds and legumes.

How is this diet information sheet different?

In 2022/2023, Global DARE Foundation funded further food analysis. This diet guide has been updated to include these new foods.

Furthermore, the diet guide is now structured to include Level 1 and Level 2 advice. We know that phytanic acid in foods directly affects phytanic

acid blood levels. However, further research is still needed to understand how phytyl fatty acid esters and free phytol are absorbed in the gut and how they affect phytanic acid levels in the body. Until we have this research, it has been decided that the Level 1 advice is based only on phytanic acid content of foods. For this reason, you will notice that there has been a change to where some foods are categorised in the diet guide. In addition, Level 1 advice includes recommendations for regular meals, carbohydrates, exercise and illness management.

Level 2 advice considers foods high in phytyl fatty acid esters that could potentially influence phytanic acid blood levels. Current food analysis does not provide conclusive free phytol content, and therefore, it has not been considered in Level 2 diet guide until we have further research.

We would recommend following all the Level 1 advice as a first line in managing your phytanic acid levels. If, after following all the Level 1 advice, you continue to have higher phytanic acid levels, we would recommend also considering the Level 2 guidance.

Level 1 guidance

It is recommended that all people with Adult Refsum disease follow the Level 1 dietary recommendations.

Please note, the guidance below provides general dietary recommendations that can be used by all individuals with Adult Refsum disease. However, everyone's food choices, portion sizes and phytanic acid levels are different, so it is recommended to speak with your dietitian to get individualised advice.

What foods can I safely eat?

These foods contain only small amounts of phytanic acid and can be eaten ***freely.***

Meats and eggs
Chicken
Chicken liver
Duck breast
Eggs
Turkey
Lean pork (cuts such as pork tenderloin and loin)
Low fat ham
Bacon (back cut or loin bacon only with fat removed)

Seafood
Clams**
Coley
Lobster*
Perch (wild)
Prawns / shrimp
Scallops
Sole

Meat alternatives
Quorn
Tofu

Legumes
Black beans
Butter beans
Chickpeas
Edamame beans
Green lentils
Kidney beans

Nuts and seeds
Almonds
Black chia seeds
Black sesame seeds

Brazil nuts
Cashew nuts
Coconut
Golden flaxseed
Hazelnuts
Macadamia nuts
Peanuts
Pecans
Pine nuts
Pistachios
Pumpkin seeds
Sunflower seeds
Tahini (white sesame seed paste)
Walnuts
White sesame seeds
Nut butters made from any of the above nuts/seeds

Milk and milk alternatives

Coconut, oat, and soya yoghurt
Fat-free cow's yoghurt
Fat free or 1-2% fat cow's milk (with no DHA)
Oat, coconut, rice and soya milk
Very-low fat fromage frais
Whey powder
Dairy free whipping cream

Cheese and cheese alternatives

Vegan and dairy-free cheese
Vegan cheese spread
Fat-free mozzarella

Cereal and cereal products

Bread (white, rye, wholemeal, wholegrain)
Breakfast cereals (with no added milk/dairy)
Crumpets
Flour (white, wholemeal)
Pasta / noodles
Pearl barley
Porridge oats
Rice
Sago
Tapioca

Fruits
All fresh and dried fruit

Vegetables
All fresh and frozen vegetables

Fats and oils
Vegetable oils
Nut and seed oils made from any of the nuts and seeds listed above (except flaxseed / linseed oil)
Vegetable suet
Vegan and vegetable oil spreads

Miscellaneous
Nori
Wasabi
Fish sauce
Herbs
Spices
Ketchup / tomato sauce
Bisto gravy powders
Stock - chicken or vegetable
Honey
Jam
Marmite
Cocoa powder
Crisps / potato chips
Ready-made sauces containing low phytanic acid ingredients (including whey protein, milk protein or skimmed milk)
Homemade puddings with only low phytanic acid ingredients

Biscuits and confectionery
Boiled sweets or sugar candy
Dairy free baking (not containing any dairy milk, butter and cream)
Dark chocolate (e.g. 70 or 90%)
Liquorice
Vegan / dairy-free chocolates
Vegan baking
Vegan ice cream

Beverages

Tea
Fruit juice
Coffee (see information on caffeine recommendations)

*There is only analysis data available for Maine lobster. The assumption is that the content is the same for other lobster; however farming practices may influence the phytanic acid content.

**There is only analysis data available for minced clams. The assumption is that the content is the same for all clams; however farming practices may influence the phytanic acid content.

CAUTION

These foods need to be ***limited*** as they contain moderate amounts of phytanic acid.

A total of a ***100g portion*** of one or any combination of the below foods can be taken ***once a day***.

If your phytanic acid levels are not responding to these restrictions (i.e., remain high), consult your dietitian for specific guidance.

Meats
Frankfurter sausage
Rabbit
Veal
Fatty bacon
Pork belly, pork lard, sausages, and shoulder (higher fat cuts)
Mortadella
Dried pork (prosciutto)
Salami

Seafood
Calamari (not coated/crumbed)
Dressed tinned crab
Halibut
Herring
Tinned tuna (in water / brine)

Legumes
Baked beans

Dairy
Fat-free and low-fat cottage cheese

Cereals
Rice crispies

STOP

These foods need to be ***avoided*** as they are high in phytanic acid.

Meats

Beef
Goat
Lamb
Mutton
Sheep
Venison
Pork offal (tongue, liver, kidney)

Seafood

Cod
Crab, tinned, white meat
Flounder
Fresh tuna
Haddock
Mackerel
Oysters
Plaice
Salmon
Sardines
Smoked salmon
Trout

Milk and milk products

All dairy creams (eg double cream, sour cream, single cream, whipping cream)
Buttermilk
Fat-free milk with added DHA (Omega 3)
Full-fat, whole or milk with more than 2g fat per 100ml
Evaporated milk
Full-fat yoghurt
Ice cream

Cheese and cheese products

Cheese spread
Full fat cottage cheese
Cream cheese

Hard and soft cheeses (cow, sheep, goat, including lite cheese)
Processed cheese
Halloumi

Cereal and cereal products
Breads with dairy (e.g. brioche, croissants)
Cereals with dairy

Vegetables
Potato flakes, dried potato or packet mash potato (if dairy / milk present in ingredients list)

Fats and oils
Algae oil
Beef suet
Butter and half-fat butter
Flaxseed / linseed oil
Fish oil
Ghee
Margarine (if it contains milk and milk products)

Miscellaneous
Stock – beef or lamb
Ready-made sauces containing dairy such as cream or milk

Biscuits and confectionery
All baking that includes butter, milk, or other dairy
Chocolate (white and milk)

Carbohydrates are important: These should be included at each meal

The brain needs a regular supply of glucose to function. Without a regular supply of glucose, your body will break down its carbohydrate stores and once it has used those, it will break down your fat stores and release phytanic acid from your body fat. It is very important **not** to follow a low carbohydrate diet. Carbohydrates can come from starchy foods (bread, potatoes, rice, pasta, breakfast cereal) and sugary foods (fruit, sweets, fruit juice). Carbohydrates should be distributed throughout the day (included at each meal and in snacks, including a bed time snack), and should not be less than 200g of carbohydrates in total per day. Carbohydrate free meals should be avoided.

If you have other health conditions such as diabetes, it is important to talk with a dietitian for specific advice.

Ensuring adequate carbohydrates when exercising

It is important to fuel your body with carbohydrates prior to exercise. This could include eating a carbohydrate containing meal or snack before exercising. If exercise lasts longer than 45 minutes, you should take a carbohydrate snack during exercise. For example, if you go on a 90-minute walk you should bring a small snack to eat at 45 minutes (e.g. banana, a couple of dates). If you exercise without adequate carbohydrates, your body will start to break down fat and release phytanic acid. If you are starting a new exercise plan or wanting to increase the amount of exercise you do, it is important to talk with your dietitian.

Protein sources for Adult Refsum disease

Protein is an important nutrient for growth, development and maintenance of muscle. Protein foods should make up one-third of your plate at meals. Some protein foods are very high in phytanic acid (e.g. beef, lamb, cheese, full-fat milk) and need to be avoided. Below is a list of protein foods that can be eaten daily. You will also get some protein from vegetables, cereals and grains. You can use the traffic light lists above for

further ideas of protein foods (including those that can be eaten in moderation on the 'caution' list).

- Chicken, turkey, duck breast, lean pork eg pork loin, ham without fat, back bacon without fat
- Eggs
- Prawns/shrimp, scallops, lobster, clams
- Perch (wild), coley, sole
- Quorn
- Tofu
- Kidney beans, black beans, green lentils, butter beans, chickpeas
- Whey powder
- Fat-free cow's yoghurt
- Soya yoghurt
- Fat-free milk
- Almonds, cashew nuts, walnuts, pistachios, hazelnuts, macadamia nuts
- Pumpkin and sunflower seeds

Other Level 1 dietary recommendations:

Eat regular meals and snacks

This means having breakfast, lunch, evening meal and a bedtime snack. If you have a long break between meals (4 hours or more), have a snack. It is particularly important to eat breakfast, so that you break your overnight fast. It is also important that you do not exercise in the morning unless you have eaten breakfast.

Dieting to lose weight is **not** recommended unless under close supervision of your medical and dietitian team. If weight loss is desired, it is important that you work closely with your medical team and dietitian to develop a weight loss plan and regularly monitor your phytanic acid levels.

Any time you lose weight phytanic acid is released from your fat stores; therefore, very slow weight loss would be recommended.

Avoid high intakes of caffeine

Consuming large amounts of caffeine from energy drinks, sports drinks, or high intake of coffee is not recommended. If consuming products with caffeine, a moderate caffeine intake is advised. For an adult woman (not intending to become pregnant) this would be 200-350mg a day and for an adult man, 300-450mg a day. Examples of caffeine content in various products are listed below:

Product	Serving Size	Caffeine Content
Sports or energy drinks	Per can or bottle	80-300mg
Mug of tea	250mls	40-70mg
Mug of instant coffee	250mls	52-85mg
Cola	330ml can	11-70mg
Pharmaceutical products	Per tablet	25-65mg
Energy supplements	Per supplement	35-50mg

Getting enough glucose when unwell

If you are feeling unwell and unable to manage all your usual meals, you will need to consume glucose at regular intervals in order to stop the breakdown of your fat stores and release of phytanic acid into your body. This is called an emergency regime, and an example of one used at Guy's and St Thomas' Hospital, London, is included (see Emergency Regimen). This regimen is specific for those who do not have diabetes. If you have diabetes, it is important to contact your medical team and dietitian for specific advice.

Medical procedures or surgery that requires fasting

Fasting for a surgical or medical procedure can result in increasing phytanic acid levels. To prevent the increase in phytanic acid levels,

glucose solutions (oral and IV) will need to be provided prior, during and post procedure. If you are having a surgery or medical procedure, please contact your medical/dietetic team to make them aware.

Ensure you are getting your essential fatty acids

Omega 3 and omega 6 are essential fatty acids that the body cannot synthesise and therefore need to be consumed in the diet. Omega 3 and 6 are found in vegetable oils, and can be a source of essential fatty acids on a low phytanic acid diet. In addition, omega 6 can also be found in chicken, eggs and wholegrain breads. It is important to discuss with your dietitian about ensuring you are getting omega 3 and 6 from your diet.

Ensure adequate vitamins and minerals

With all specialist diets it is important to regularly monitor nutritional bloods (for example, vitamin B12, folate, ferritin, vitamin D). These should be monitored annually and vitamin / mineral replacement provided as needed.

Level 2 guidance

If phytanic acid levels continue to be high despite following **all** Level 1 advice, we would recommend reviewing your diet to see if any of the below foods feature. These foods contain phytyl fatty acid esters, which could be contributing to your phytanic acid levels in your blood. If you are consuming any of these foods regularly, it is recommended to speak with your dietitian for specific guidance.

CAUTION

These foods contain **moderate amounts** of phytyl fatty acid esters.

A total of a ***100g portion*** of one or any combination of the below foods can be taken ***once a day***.

Tahini (white sesame seed paste)
White sesame seeds
Pumpkin seeds
Sesame seed oil
Olive oil
Soy oil
Peanut oil
Peanut butter
Peanuts
Pecans
Broccoli
Rocket / arugula
Red and yellow bell peppers
Green olives
Edamame beans
Chickpeas
Dairy free / vegan butters, cheese, cream cheese, cheese spread or whipping creams **containing any of the above oils**

STOP

These foods are **high** in phytyl fatty acid esters.

Flaxseed / linseed oil
Black sesame seeds
Sunflower seeds
Golden flaxseed
Kale
Soy burger (Beyond beef)
Dark chocolate
Dairy free / vegan butters, cheese, cream cheese, cheese spread or whipping creams **containing flaxseed / linseed oil**

Meal swap examples

Level 1 guidance

Croissant, butter and coffee

× High phytanic acid foods

Wholemeal bread, vegan spread, eggs, orange juice

✓ Low phytanic acid foods

Description: A breakfast with croissant, butter and coffee that would be high in phytanic acid. This can be swapped for two slices of wholemeal bread with vegan spread, eggs and glass of orange juice as a breakfast lower in phytanic acid.

Level 1 guidance

1 x bread, vegan spread and tea

× Not enough calories

2 x bread, vegan spread, soya yoghurt, banana and tea

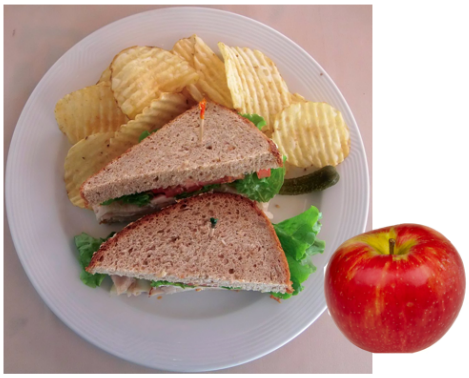
✓ Adequate calorie

Description: A meal of one slices of bread with vegan spread and cup of tea. This meal would not contain enough calories. To ensure adequate calories the meal could be swapped for two slices of bread with vegan spread, along with soya yoghurt, a banana and tea.

Level 1 and 2 guidance

Chicken, rocket/arugula, carrots, apple

× Low in carbohydrates
× Moderately high in phytyl esters



Chicken salad sandwich (wholemeal bread, lettuce, vegan spread, tomato), crisps and apple

✓ Carbohydrate portion
✓ Low in phytyl esters

Description: A lunch of grilled chicken, rocket/arugula, carrots salad and an apple. This lunch would be too low in carbohydrates (Level 1 advice) and contain foods moderately high in phytyl esters (Level 2 advice). This can be swapped for a chicken salad sandwich, crisps and an apple, which would contain a carbohydrate portion and be lower in phytyl esters.

Emergency regime

Example of regimen used at Guy's and St Thomas Hospital, London

The emergency regime (ER) of glucose polymer drinks should be commenced immediately if you become unwell, e.g. nausea, vomiting, diarrhoea, high temperature, or any illness resulting in loss of appetite and inability to take your normal diet.

The ER is used to prevent the release phytanic acid from your body fat which can occur if your body is not getting adequate carbohydrates during illness. It is important to have the right amount of glucose to maintain your body metabolism, and prevent breaking down of your body's alternative source of energy during this time.

Stage 1

At the first sign of feeling unwell and loss of appetite, take 200 ml glucose polymer drink **(see recipes on the following page)**. If you feel better within one to two hours, you may return to your normal diet.

Stage 2

If you continue to feel unwell, commence the full emergency regime as described below and contact the Inherited Metabolic team.

Stage 3

If you are unable to tolerate the emergency regime, if you do not improve, or if you become increasingly unwell, go to your local Accident and Emergency (A&E) and ask the hospital healthcare team to contact the Inherited Metabolic team.

The full emergency regime

- Take the ER drinks every two hours **day and night**
- If tolerated, continue all prescribed medicines

- You should not use your ER for more than 2 days without your normal diet. If your symptoms persist and you are unable to tolerate your ER, contact your GP or go to your nearest Accident and Emergency (A&E). Inform your Inherited Metabolic team.
- Continue with extra glucose polymer drinks during the day until the normal diet is resumed

Recipes for 20% emergency regimen drink

One of the following recipes will be recommended by your dietitian. These can be made up using 200ml of water. Consume all 200ml of the ER drink every 2 hours day and night.

- 40g Maxijul Super Soluble, Polycal or Vitajoule powder
- OR 1 sachet of S.O.S 20 (Vitaflo)

Alternative oral emergency drinks for adults

Ideally 200ml of the emergency drink (as per recipe above) must be taken every 2 hours when you are unwell. If you cannot take any of the ER drink or you don't have access to them quickly, then you can start with an alternative drink such as the options given below. You will need to drink more than 200ml of the alternative drinks every 2 hours to get your carbohydrate needs.

Mountain Dew Citrus Burst (13g carbohydrates per 100ml)	Take **300ml** every 2 hours
Coca Cola (full sugar) (10.6g carbohydrates per 100ml)	Take **380ml** every 2 hours
Pepsi (full sugar) (11g carbohydrates per 100ml)	Take **380ml** every 2 hours
Fruit juice (eg apple juice) with at least 10g carbohydrates per 100ml	Take **400ml** every 2 hours

Note:

- Carbohydrate content of the drinks can change. Please regularly check the carbohydrate content per 100ml to ensure it still contains the above amount of carbohydrate.
- Some of the above alternative drinks contain caffeine. Use of these for 24 hours during illness would be appropriate. When well, to consume in moderation.

Contact Details

St Thomas' Hospital Adult Inherited Metabolic Service

Consultant's Secretary:	gst-tr.adultmetabolic@nhs.net Monday-Friday (0900-1700) Tel: +44 (0) 20 7188 4004
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Patient Support Group

Global DARE Foundation

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. DARE stands for Defeat Adult Refsum Everywhere.

At DARE's website there are educational resources including recorded webinars on Refsum disease and this diet.

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