

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. The levels of this fat build up in the body over time and eventually cause symptoms.

The symptoms of a high phytanic acid level in the blood are 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.

Lifespan is normal. Children of a person with Adult Refsum Disease are unlikely to be affected because the parent who does not have Adult Refsum's Disease is unlikely to be a carrier for the gene.

How does the Westminster Refsum's Diet work?

The diet works in two ways:

1. It restricts the intake of foods which are rich in phytanic acid and the foods which the body can convert to phytanic acid to a level that the body can process.
2. It supports general metabolism so that the body does not release phytanic acid from body stores in amounts that are greater than the body can process.

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 a fatty acid that is found in small amounts in many foods. Dietary advice aims to avoid the foods which contain a large amount of phytanic acid.

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: Phytol 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 2021/2022, Global DARE Foundation funded the analysis of 60 new foods. Based on this analysis, we have learnt more about the phytanic acid, phytyl fatty acid esters and free phytol content of foods. With having more information about foods, we have been able to provide a traffic light system for the dietary advice. The food analysis methods have changed over the years and this, along with having more information about different foods has led to some big changes to the dietary guidance information.

Further research is still needed to understand how phytyl fatty acid esters and free phytol are absorbed in the gut. Until we have this research, advice is based on the content of these substances present in foods.

NEW Traffic Light System

GO	These foods contain only small amounts of phytanic acid, phytyl fatty acid esters and free phytol and can be eaten freely .
CAUTION	These foods need to be limited to a total of a 100g portion of one or any combination of foods once a day as they contain moderate amounts of phytanic acid, phytyl fatty acid esters and free phytol.
STOP	These foods need to be avoided as they are high in phytanic acid, phytyl fatty acid esters or free phytol

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.

GO	These foods contain only small amounts of phytanic acid, phytyl fatty acid esters and free phytol and can be eaten freely .
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Meats and eggs	Fruits
Chicken	All fresh and dried fruit
Turkey	
Duck breast	Vegetables
Rabbit	Sweet peas
Eggs	Sweet potato
Chicken liver	Asparagus
	Brussel sprouts
Seafood	Broad beans
Scallops	Bean shoots
Lobster*	Lettuce (iceberg)
Clams**	Peas
Perch (wild)	Cabbage
	Carrots
Meat alternatives	Onions
Quorn	Cucumber
Tofu	Sauerkraut
	Butternut squash
Legumes	Avocado
Kidney beans	Green beans
Black beans	Cauliflower
Green lentils	Potatoes (including fries)
Butter beans	Beetroot
	Mushrooms
Milk and milk alternatives	Swede
Fat free or 1% fat cow’s milk (with no DHA)	Eggplant / Aubergine Green peppers
Oat, coconut, rice and soya milk	Tomatoes (fresh, canned, puree)

Coconut, oat, and soya yoghurt	Cheese alternatives
Whey powder	Vegan and 'dairy free' cheeses
Very low fat Fromage Frais	Vegan cheese spreads
Cereal and cereal products	Nuts and seeds
Bread (white, rye, wholemeal, wholegrain)	Cashew nuts Walnuts
Crumpets	Coconut
Cereals (cornflakes, weetabix, shredded wheat, bran flakes, crunchy nut, cocopops, honey nut loops, Kelloggs Special K, puffed wheat cereal)	Pistachios Hazelnuts Black Chia seeds Macadamia nuts
Porridge oats	Nut butters made from any of the above nuts/seeds
Flour (white, wholemeal)	
Rice	Miscellaneous
Pasta / noodles	Nori
Sago	Wasabi
Tapioca	Fish sauce
Pearl barley	Herbs
	Spices
Fats and oils	Ketchup / tomato sauce
Vegetable oils	Bisto gravy powders
Vegetable suet	Stock - chicken or vegetable
Vegan and vegetable oil spreads	Honey
	Jam
Biscuits and confectionery	Marmite
Vegan baking (not containing any of the nuts and seeds that are on the avoid list)	Crisps / potato chips
Dairy free baking (not containing any dairy milk, butter and cream)	Ready-made sauces containing GO ingredients (including whey protein, milk protein or skimmed milk)
Vegan/dairy free chocolates Dark chocolate (e.g. 70 or 90%) Boiled sweets or sugar candy	
Liquorice	Beverages
	Tea
	Fruit juice

	Coffee (see information on caffeine recommendations)
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*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, phytyl fatty acid esters and free phytol.

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 (ie remain high), consult your Dietitian for specific guidance.

Meats	Vegetables
Veal	Broccoli
Pork	Spinach
Ham	Rocket / Arugula
Bacon	Sweetcorn
Frankfurter sausage	Red and yellow bell peppers
	Olives
Seafood	Nuts and seeds
Tinned tuna (water/brine)	White sesame seeds
Halibut	Pumpkin seeds
Calamari (not coated/crumbed)	Pecans
Coley	Tahini (white sesame seed paste)
Herring	
Sole	Legumes
Dressed tinned crab	Edamame beans
Prawns / Shrimp	Chickpeas
	Baked beans
Dairy	
Fat-free cow's yoghurt	Cereals
	Rice crispies

STOP

These foods need to be **avoided** as they are high in phytanic acid, phytol fatty acid esters or free phytol

Meats	Vegetables
Cattle (steak, beef, burgers)	Kale
Sheep Mutton Lamb	Potato flakes, dried potato or packet mash potato (if dairy/milk present in ingredients list)
Goat	
Beef, sheep, lamb and pork offal	Nuts and seeds
Dried pork (prosciutto)	Black sesame seeds
	Sunflower seeds
Seafood	Golden Flaxseed
Fresh tuna or tinned in oil	Peanuts
Trout	Peanut butter
Oysters	Almonds
Smoked salmon	Brazil nuts
Salmon	
Cod	Fats and oils
Flounder Haddock	Margarine (if it contains milk and milk products)
Plaice	Butter and half-fat butter
Crab, tinned, white meat	Beef suet
Mackerel	Ghee
Sardines	Fish oil
	Algae oil
Milk and milk products	Peanut oil
Full fat, whole or semi-skimmed milk Evaporated milk	Sesame seed oil
Fat free milk with added DHA (Omega 3)	
Full fat yoghurt	Cheese and cheese products
All dairy creams (eg double cream, sour cream, single cream, whipping cream)	Hard and soft cheeses (cow, sheep, goat)
Ice cream	Processed cheese Cream cheese
Buttermilk	Cottage cheese
	Cheese spread
Cereal and cereal products	

Breads with dairy (e.g. brioche, croissants)	
Cereals with dairy	
	Miscellaneous
Biscuits and confectionery	Stock – beef or lamb
All baking that includes butter, milk or other dairy	Ready-made sauces containing dairy such as cream or milk
Chocolate	

Protein sources for Adult Refsum Diseases

Protein is an important nutrient for growth, development and maintenance of muscle. Protein foods should make up 1/3 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, rabbit
- Eggs
- Scallops, lobster, clams
- Perch (wild)
- Quorn
- Tofu
- Kidney beans, black beans, green lentils, butter beans
- Whey powder
- Very low fat Fromage Frais
- Soya yoghurt
- Fat-free milk
- Cashew nuts, walnuts, pistachios, hazelnuts, macadamia nuts

The Westminster Refsum's Diet

The Westminster Refsum's Diet has been shown to be effective at lowering the amount of phytanic acid in the blood. The Westminster Refsum's Diet includes the following principles:

1. 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 hard in the morning unless you have eaten breakfast.

Dieting to lose weight is **not** recommended unless your phytanic acid is in the target range of less than 200µmol/L. 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.

2. Eat carbohydrate rich foods 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).

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

3. Avoid foods that contain a lot of phytanic acid, phytyl fatty acid esters and free phytol

See **STOP** section for foods to avoid

4. Limit foods that contain a moderate amount of phytanic acid, phytyl fatty acid esters and free phytol

See **CAUTION** section for foods to limit.

5. 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
Chocolate bar	50g bar	5-36mg
Pharmaceutical products	Per tablet	25-65mg
Energy supplements	Per supplement	35-50mg

6. 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 walnut oil, 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.

7. Ensure adequate vitamins and minerals

With all specialist diets it is important to regularly monitor nutritional bloods. These should be monitored annually and vitamin / mineral replacement provided as needed.

8. 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.

9. 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.

10. Medical procedures or surgery that requires fasting

Fasting for a surgical or medical procedures 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.

Example Meal Plan

Meal/time	Food suggestions
Breakfast	<p>Fruit or fruit juice, tea or coffee</p> <p>Breakfast cereal with soya milk or fat free cow's milk</p> <p>Any bread (except those that contain dairy such as brioche or cholla) with dairy free spread and jam, marmalade, honey or marmite</p> <p>Eggs, chicken sausages/vegetarian sausages, tomatoes, mushrooms and bread</p> <p>Dairy free croissants</p>
Mid-morning	<p>Fruit</p> <p>Dairy free biscuits or cake or cereal bar</p> <p>Soya, oat or coconut yoghurt</p>
Lunch	<p>Meal to comprise (can be as a sandwich or cooked meal):</p> <p>Dairy free spread or oil to be used as a spread or in cooking or as a dressing on salad.</p> <p>Protein:</p> <p>Options could include: Chicken, eggs, pulses or beans, quorn, tofu, dairy free cheese, rabbit, turkey</p> <p>Vegetables or Salad (from the GO list)</p> <p>Carbohydrate:</p> <p>Options could include: Bread, bread roll, pitta, wrap, jacket potato, pasta, rice, sweet potato, noodles</p> <p>Dairy free chocolate bar/dairy free dessert</p> <p>Fresh fruit</p>
Mid-afternoon	<p>Fresh fruit / vegetable sticks / dairy free cereal bar / dairy free biscuits or cake, crisps (potato chips), popcorn / dairy free chocolate bar / soya, coconut, or oat yoghurt</p>

Meal/time	Food suggestions
Evening meal	<p>Meal to comprise (can be as a sandwich or cooked meal):</p> <p>Dairy free spread or oil to be used as a spread or in cooking or as a dressing on salad.</p> <p>Protein:</p> <p>Options could include: Chicken, eggs, pulses or beans, quorn, tofu, dairy free cheese, rabbit, turkey</p> <p>Vegetables or Salad (from the GO list)</p> <p>Carbohydrate:</p> <p>Options could include: Bread, bread roll, pitta, wrap, jacket potato, pasta, rice, sweet potato, noodles</p> <p>Dairy free chocolate bar/dairy free dessert</p> <p>Fresh fruit</p>
Evening Snack	<p>Dairy free biscuits / dairy free cake / breakfast cereal with soya milk or fat free cow's milk / bread with dairy free spread</p>
Fluids	<p>Consume fluids throughout the day such as water, tea, herbal teas, coffee, fat free cow's milk or milk alternatives</p> <p>Note: Caffeine containing drinks should be limited to no more than 3 a day</p>

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 200mls 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 200mls 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 200mls 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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