

# Understanding Adult Refsum's Disease

## Diet Information Sheet

### What is Adult Refsum's Disease?

Adult Refsum's 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, difficulty in coordinating walking and balance, the skin becomes itchy and scaly as well as other symptoms.

### What is the treatment for Adult Refsum's 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's Disease are unlikely to be affected because the parent who does not have Adult Refsum's Disease would have 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.

## Which foods contain phytanic acid?

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) and when bacterial cultures are added to foods such as in the production of yoghurt or kimchi.

All the information provided below is based on the foods that have been analysed for phytanic acid content in the past and clinical experience. The variety of foods and farming practices have changed significantly since the prior testing. Therefore, further funding is being sought to

repeat previous analyses and analyse new foods to improve upon this guidance.

## Meat and dairy

The foods with the highest phytanic acid content are the meat and dairy products from cattle, sheep, lambs and goats. Organic products from these animals contain significantly more phytanic acid than the non-organic versions due to being grass fed.

### Recommendation:

- Choose chicken, turkey, pork and ham instead.
- Soya meat alternatives or tofu can be used.

The liver of cattle, sheep, lambs and pigs is a rich source of phytanic acid.

### Recommendation:

- Choose chicken liver instead.

Dairy products like butter, cheese, milk, yoghurt and cream from cows, sheep or goats are rich in phytanic acid.

### Recommendation:

- Choose fat free milk (skimmed milk) – this should be limited to 150ml daily.
- Non-dairy products made using soya, oats, rice and coconut would be good alternatives. Any nut-based products should be limited to 30g per week.
- Fat free quark and fat free fromage frais are both skimmed milk cheeses and can be eaten unless they contain added cream. The portion size for these products should be limited to 50g daily.

- Fat free yoghurts should be avoided because the way they are made causes the traces of fat they contain to be converted to phytanic acid.

## Seafood

Oily fish and large fish such as tuna are also rich sources of phytanic acid.

### Recommendation:

- Choose cod, coley, haddock, crab and prawns instead.

## Other Products

Cereal products, like breakfast cereals, rice, quinoa and pasta do not contain significant amounts of phytanic acid.

Most cooked and raw vegetables, salad and fruit are low in phytanic acid. See below for recommendations of some vegetables which contain substances that are converted to phytanic acid and need to be limited.

A quick reference table of foods with high, moderate, and low phytanic acid content is provided at the end of this document

## Which foods contain substances that are converted to phytanic acid by the body?

Two substances can be converted to phytanic acid by the body.



Both of these substances come from vegetables and fruit. Diets heavily based on plants will have less predictable effects on the phytanic acid concentration in the blood because we have less information about these foods.

Phytyl fatty acid esters are substances that are found in plants. Some recently published research demonstrated that these substances can be converted into phytanic acid. We hope to analyse a broader range of fruit and vegetables and learn more about these substances.

### **Foods rich in phytyl fatty acid esters**

Phytyl fatty acid esters have been found in: red and yellow bell peppers, rocket salad (arugula), green olives, red grapes, carrots and cucumber.

Eating small amounts of these foods is unlikely to make a significant difference to the amount of phytanic acid in the blood, however, we would advise you limit red or yellow pepper (eat green ones instead), and rocket salad to 50g per day. The quantities of green olives, cucumber, red grapes and carrots that would need to be eaten make it very unlikely that they would make a significant difference to the amount of phytanic acid in the blood. However, if eating olives, black olives contain no phytyl fatty acid esters.

### **Foods rich in free phytol**

Free phytol is found in many animal and vegetable products.

Previous studies suggested that free phytol contributes less than 10% of the phytanic acid in the blood. Further information is required before advice can be given.

## 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 and your phytanic acid is low, it is important that you work closely with your 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**

The foods that contain the most phytanic acid are products from:

- Cows (milk, butter, cheese, cream, products that contain these substances such as cakes or biscuits; ice-cream, beef, burgers, sausages, and meat balls containing beef)
- Sheep (mutton, lamb, sheep's cheese, sheep's milk, sheep's yoghurt)
- Oil rich fish (salmon, mackerel, herring, sardines, pilchards, trout, mullet, sea bass, fish oil supplements).
- Organic versions of these foods and foods with a very high fat content contain the most phytanic acid e.g. organic butter, organic cheddar, fish oil.

### **4. Restrict foods that contain a moderate amount of phytanic acid to a portion size not bigger than 80g and do not eat them more than one a week**

The foods that contain a moderate amount of phytanic acid are duck and tinned dressed crab. Nuts should be restricted to less than 30g as a portion and limited to once a week.

### **5. Limit foods containing phytyl fatty acid esters to 50g a day**

These foods are red and yellow peppers (green peppers do not need to be limited) and rocket salad (arugula).

### **6. 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:

<b>Product</b>	<b>Serving Size</b>	<b>Caffeine Content</b>
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

## **7. 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 plant oils such as flaxseed (linseed), soybean, and canola 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.

## **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.

## Recommended Meal Plan

Meal/time	Food suggestions
<b>Breakfast</b>	<p style="text-align: center;">Fruit or fruit juice</p> <p style="text-align: center;">Breakfast cereal with soya milk</p> <p style="text-align: center;">Any bread (except those that contain dairy such as brioche or cholla) with dairy free spread and jam, marmalade, honey or marmite</p> <p style="text-align: center;">Eggs, bacon, pork sausages/vegetarian sausages, beans and bread</p> <p style="text-align: center;">Dairy free croissants</p>
<b>Mid-morning</b>	<p style="text-align: center;">Fruit</p> <p style="text-align: center;">Dairy free biscuits or cake or cereal bar</p> <p style="text-align: center;">Soya yoghurt</p>
<b>Lunch</b>	<p>Meal to comprise (can be as a sandwich or cooked meal):</p> <p style="text-align: center;">Dairy free spread or oil to be used as a spread or in cooking or as a dressing on salad.</p> <p style="text-align: center;">Protein:</p> <p style="text-align: center;">Options could include: Chicken, eggs, dairy free cheese, falafel, pulses or beans, hummus, cod, coley, haddock, prawns, rabbit, turkey or pork</p> <p style="text-align: center;">Vegetables or Salad</p> <p style="text-align: center;">Carbohydrate:</p> <p style="text-align: center;">Options could include: Bread, bread roll, pitta, wrap, jacket potato, pasta, rice, sweet potato, noodles</p> <p style="text-align: center;">Dairy free chocolate bar/dairy free dessert</p> <p style="text-align: center;">Fresh fruit (e.g. a banana or a pear or an apple)</p>
<b>Mid-afternoon</b>	<p style="text-align: center;">Fresh fruit / carrot sticks and hummus/ dairy free cereal bar/dairy free biscuits or cake/crisps/popcorn/ dairy free chocolate bar/soya yoghurt</p>

<b>Meal/time</b>	<b>Food suggestions</b>
<b>Evening meal</b>	<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, dairy free cheese, falafel, pulses or beans, hummus, cod, coley, haddock, prawns, rabbit, turkey or pork</p> <p>Vegetables or Salad</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 (e.g. a banana or a pear or an apple)</p>
<b>Evening Snack</b>	<p>Dairy free biscuits/dairy free cake/breakfast cereal with soya milk/ bread with dairy free spread</p>
<b>Fluids</b>	<p>Consume fluids throughout the day such as water, tea, herbal teas, coffee and soya milk</p> <p>Note: Skimmed milk to be limited to 150ml daily. 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 Adult 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 Adult 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.

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

<b>Consultant's Secretary:</b>	<a href="mailto:gst-tr.adultmetabolic@nhs.net">gst-tr.adultmetabolic@nhs.net</a> Monday-Friday (0900-1700) Tel: +44 (0) 20 7188 4004
<b>Adult Inherited Metabolic Dietitians</b>	<a href="mailto:gst-tr.adultimddietitians@nhs.net">gst-tr.adultimddietitians@nhs.net</a> Monday-Friday (0900-1700) Tel: +44 (0) 20 7188 9611
<b>Adult Inherited Metabolic Nurse</b>	<a href="mailto:gst-tr.adultimdnurse@nhs.net">gst-tr.adultimdnurse@nhs.net</a> Monday-Friday (0900-1700) Tel: +44 (0) 20 7188 0850

## 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.

<b>Primary Contact</b>	Kristie DeMarco kristiedemarco@globaldarefoundation.org" <a href="mailto:kristiedemarco@globaldarefoundation.org">kristiedemarco@globaldarefoundation.org</a>
<b>Website</b>	<a href="https://www.defeatadultrefsumeverywhere.org/">https://www.defeatadultrefsumeverywhere.org/</a> <a href="https://www.defeatadultrefsumeverywhere.org/">https://www.defeatadultrefsumeverywhere.org/</a>
<b>Refsum Community Connections</b>	<a href="https://www.defeatadultrefsumeverywhere.org/refsum-community-connection">https://www.defeatadultrefsumeverywhere.org/refsum-community-connection</a>

## Food Table – Quick Reference Guide

Foods to Avoid (High Phytanic Acid)	Foods to Limit (Moderate Phytanic Acid)	Foods to Eat (Low in Phytanic Acid)
<b>Meat Products</b>		
Cattle (steak, beef, burgers) Sheep Mutton Lamb Goat Sausage containing beef Meatballs containing beef Cattle liver Sheep liver Lamb liver Pigs liver	Duck/goose – limit to 80g per week	Chicken Turkey Pork Bacon Pork sausages Ham Soya meat alternatives Tofu Eggs Chicken Liver
<b>Seafood</b>		
Salmon Mackerel Herring Sardines Pilchards Trout Mullet Sea bass Fish oil supplements	Tinned Dressed Crab – limit to 80g per week	Cod Coley Haddock Crab Prawns
<b>Vegetables / Salad / Fruit</b>		
	Limit these to 50g per day: <ul style="list-style-type: none"> <li>• Red Pepper</li> <li>• Yellow Pepper</li> <li>• Rocket Salad (Arugula)</li> </ul>	All other raw and cooked vegetables, legumes, salad, fruit are low in phytanic acid.



Foods to Avoid (High Phytanic Acid)	Foods to Limit (Moderate Phytanic Acid)	Foods to Eat (Low in Phytanic Acid)
<b>Dairy Products</b>		
<p>Dairy products from cows, sheep, goat such as:</p> <ul style="list-style-type: none"> <li>• Cheese</li> <li>• Milk</li> <li>• Cream</li> <li>• Yoghurt including fat free</li> <li>• Butter</li> <li>• Ghee</li> <li>• Ice Cream</li> </ul>	<p>Fat free milk (skimmed milk) – limit to 150ml daily</p> <p>Fat free quark and fat free fromage frais can be eaten unless they contain added cream. These products should be limited to 50g day</p>	<p>Non-dairy products made using soya, oats, rice and coconut would be good alternatives.</p> <p>Any nut-based products should be limited to 30g per week.</p>
<b>Other Foods</b>		
<p>Breads with dairy (e.g. brioche, croissants)</p> <p>Cereals with dairy</p> <p>Confectionary with dairy (e.g. milk chocolate, toffee, fudge)</p>	<p>Nuts and nut-based products (e.g. nut butters) should be limited to a total of 30g per week</p>	<p>Breads without dairy</p> <p>Cereals without dairy</p> <p>Confectionary without dairy (e.g. dark chocolate, carob, chocolate that doesn't contain milk)</p> <p>Rice</p> <p>Pasta</p> <p>Quinoa</p> <p>Seeds (e.g. sunflower, pumpkin, chia)</p> <p>Oils: Vegetable oil, corn oil, olive oil, sunflower oil, safflower oil, canola oil</p> <p>Honey, Maple Syrup</p> <p>Jelly</p> <p>Tea, coffee</p>