

Understanding the CPT 1A Variant

A guide for First Nations People

What is The CPT 1A variant?

Energy from fat keeps us going whenever our bodies run low of our main source of energy, a type of sugar called **glucose**. Our bodies rely on fat when we don't eat for a stretch of time – like when we miss a meal or when we sleep. CPT 1A is an important enzyme that changes fats into energy at these times.

When the CPT 1A enzyme is not working properly, the body cannot use fat for energy as well as it should, and must rely more on glucose. Although glucose is a good source of energy, there is a limited amount available in our body. Once the glucose has been used up, the body tries to use fat for energy without much success. This can lead to low blood sugar, called **hypoglycemia**, and to the build up of harmful substances in the blood.

Symptoms of low blood sugar				
• Shaking	• Sweatiness	• Anxiety	• Dizziness	• Hunger
• Fast heartbeat	• Visual impairment	• Weakness, fatigue	• Headache	• Irritability

How do people get the CPT 1A variant?

Approximately 10% of all First Nations babies born in British Columbia each year are identified with the CPT 1A variant (Sinclair, 2011). It is most commonly found in the First Nations people who originate from Vancouver Island and Coastal regions of BC. The CPT 1A variant is a genetic change in an enzyme in the body that a person has from the time they are conceived. A baby is made up of two sets of genes, one from the mother, and one from the father.

These genes from mom and dad come together to form a unique recipe that determines the baby's eye and hair colour, sex and even whether the baby will be right or left-handed. Each pair of genes gives directions to a certain part of the body.

The CPT 1A variant comes from getting two copies of the gene that causes the CPT 1A variant, one from the mother and one from the father. If the child inherits only one copy of the gene, they carry the variant but do not have any symptoms. There is a 1 in 4, or 25% chance that 2 carriers of the gene will have a baby who has the CPT 1A variant. Boys and girls are equally affected. If a member of your family has been identified with the CPT 1A variant, then talk to your doctor about whether or not you should be tested. Your doctor can talk to you about your risk of having the CPT 1A variant.

How is the CPT 1A variant identified?

Adults are identified with the CPT I variant through a simple blood test.

What symptoms can be related to the CPT 1A variant in adults?

It is important to understand that simply having the CPT 1A variant is not enough to result in symptoms in adults. Adults who have the CPT 1A variant may be at risk of having some symptoms related to the variant if they develop another health condition as discussed below. At the current time, we do not have any proof that the CPT 1A variant can cause symptoms in adults at all. The CPT 1A variant is very common in people who are of First Nations' descent and yet symptoms which might be related to the variant are extremely rare. Therefore, it stands to reason that almost all adults who have the CPT 1A variant will NEVER have symptoms related to the variant and that the CPT 1A variant will not affect their health at all. However, there are some situations where the body is more dependent on the use of fat as an energy source and it is possible that adults with the CPT 1A variant may be at risk of symptoms during these situations. More research is needed before we know if this statement is true.

Some of the situations where it is possible that adults with the CPT 1A variant may be at risk of symptoms include:

1. Pregnancy

Women who are pregnant break down more fat than women who are not pregnant. This increase in fat break down is due to the increased hormones of pregnancy and also due to the fact that the mother is providing nutrients to the fetus. You should talk to your doctor if you develop any of the following symptoms during pregnancy:

- Excessive vomiting (throwing up)
- Abdominal pain
- High blood pressure
- Jaundice (yellow tinge to skin, whites of eyes)
- Abnormal fat storage in the liver
- Severe bleeding

2. Fasting

When your body goes without food, it uses fat as an energy source. Common situations where adults may go without food, or "fast", include:

- Fasting before a planned surgery
- Fasting due to a stomach bug such as the stomach flu
- Fasting for a cultural ceremony

Adults with the CPT 1A variant who fast for many hours may be at risk of developing low blood sugar which may present with the following symptoms:

- Extreme sleepiness
- Behavior changes
- Irritable mood
- Poor appetite

If you have been identified with the CPT 1A variant, you should talk to your doctor before undertaking any fast that lasts for more than several hours. Most adults with the CPT 1A variant should tolerate fasting overnight as is common if your doctor has ordered fasting blood tests. However, if the fasting is longer than just overnight, you should talk to your doctor about this. Your doctor may choose to give you some intravenous or I.V. sugar water while you are fasting to avoid any problems.

3. Other situations which may be associated with a risk of symptoms in adults with the CPT 1A variant:

- Long periods of exercise, including berry picking, participating in other traditional activities or ceremonies.
- Long periods of having nothing to eat
- Very hot or very cold temperatures
- Viral infections such as colds or flus
- Bacterial infections
- Anesthetic for surgery
- Lack of sleep
- High fat diet
- Having more than one serving of alcohol

What should my doctor know about the CPT 1A variant?

Let your doctor know that you have been identified with the CPT 1A variant, and that more research is needed before we will know if adults with the CPT 1A variant have symptoms related to it. Your doctor may test you to see if your blood sugars are normal. If you are planning to have surgery, your doctor may run an intravenous (IV) with some sugar water while you are fasting for your surgery and may also talk to the anesthetist about the use of some medications which may be better than others in adults with the CPT 1A variant. Your doctor can contact the Adult Metabolic Diseases Clinic at Vancouver General Hospital (604-875-5965) for further information.

Can I do anything to reduce my risk of having symptoms related to the CPT 1A variant?

Yes, you can. Through very simple dietary recommendations shared below, you can prevent your body from relying on fat as an energy source. These dietary recommendations are part of a healthy traditional diet.

There are four diet recommendations.

- Fuel your body so that the fat breakdown system doesn't kick in by eating a diet higher in carbohydrates and preventing long periods of fasting (not eating)
- Limit fatty foods that the body can not use for energy, e.g. french fries, mayonnaise, donuts, chips and deep fried foods
- Stay hydrated by drinking lots of fluids
- If you drink alcohol, limit intake to 1 serving at a time and avoid drinking on an empty stomach

We welcome collaboration with traditional healers and would be happy to work with you to include traditional medicines and foods, as well as traditional medicine practices that may involve long periods without eating into your treatment plan:

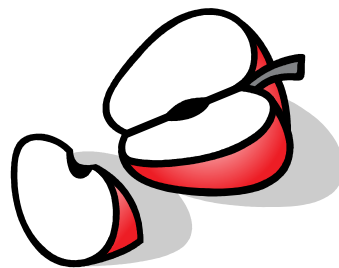
1. Fuel your body

Choose carbohydrate-rich foods that are most similar to carbohydrate containing traditional foods. These foods provide your body with glucose, so it doesn't start to break down fats for fuel:

Carbohydrate-rich food ideas	
Fruit and fruit cups	Oatmeal
Vegetables such as yams, beets, radishes, onions, turnips, parsnips	All-bran cereal, High fiber cereals
Lentils, chick peas, beans	Pasta, orzo
Bagel, pita bread, rice cakes	Corn
V8 Juice	Potatoes
Whole wheat bread	Stone ground or whole wheat crackers
Brown rice	Cous cous, quinoa
Graham crackers	Oven-baked bannock

Eat three meals a day plus 1-2 snacks. Have a nutritious snack before exercising, during prolonged exercise and before bed. Snack ideas include:

- A piece of fruit, applesauce
- Crackers and low fat cheese/low fat cream cheese
- Graham crackers and a glass skim milk
- A Fruit/Vegetable Bar (e.g. Sunrype)
- Pretzels (unsalted)
- Low fat granola bar
- A handful of your favorite dry cereal
- Oven-baked bannock
- Whole wheat bread



The balancing act

Although we are recommending you eat complex carbohydrates we want you to avoid unnecessary weight gain. It is important to choose your carbohydrates carefully. Choose those that are most like the ones available traditionally, that is, high in fiber and low in sugar and refined ingredients. By choosing complex carbohydrates that have a higher amount of fiber, you will feel fuller than when you eat processed low fiber foods.

Foods to choose less often	Try this instead....
White bread	100% stone ground whole wheat bread, Pumpernickel bread, Sprouted grain bread
Breakfast cereals with a lot of sugar and whose main ingredient is white flour	All Bran™, Bran Buds with Psyllium™, Oat Bran™, Large flake oats, Red river
	Lentils, Chickpeas, Kidney beans
White rice	Parboiled, converted or brown rice, barley, bulgur, buckwheat
Regular pasta	Whole wheat or multigrain pasta
Instant mashed potatoes	Sweet potato, yams, taro, new potato
Foods whose first ingredient is white flour	Look for food labels that state the following: 'high source of fiber', 'Made with whole grains' or whose first ingredient is whole wheat flour

2. Limit the fat in your diet

Food to choose less often	Try this instead...
Foods high in hidden fat like croissants, donuts, danishes, regular cheddar cheese, crackers (bacon dippers, cheese Ritz), cream soup, store bought muffins	<ul style="list-style-type: none"> Choose packaged foods with a low % daily value (DV) of fat on the nutrition label Low fat cheese (less than 20% milk fat) Baked crackers Home made muffins Lower fat canned soups Low fat granola
Deep-fried foods (e.g. deep-fried or pan-fried bannock, fish and chips, french fries, potato chips)	Oven-baked bannock, baked chips, home made fries, baked fish, dried fish (without oil)
Full fat mayonnaise	Light/low fat/ultra low fat mayonnaise
Creamy salad dressing	Vinaigrettes (oil & balsamic vinegar); light salad dressings
Foods to limit	How much is OK to have?
Margarine, butter, gravy, sour cream, salad dressing, oil	3-4 tsp daily in total of these foods
Eggs (each egg yolk has 5 g fat)	3 per week. Try Egg Beaters or Simply Egg whites as a substitute
Other tips:	
Trim visible fat from meats; remove skin from chicken	
Cook meat so the fat drips away (grill on rack, BBQ, rotisserie)	

Here is the nutritional content of some foods we have been asked about:

Food	Calories	Protein (g)	Carbs (g)	Fat (g)
Dried Alaskan Salmon (3 oz)	321	52	0	12
Cod, dried and salted (3 oz)	247	53	0	2
Seal, bearded, dried (3 oz)	298	70	0	2
Seal, bearded, raw (3 oz)	94	23	0	0.3
Roe (fish eggs) mixed species –baked or boiled (1 oz)	57	8	0.5	2.3
Cornstarch (1 tbsp)	30	0	7.3	0

3. Drink plenty of water

Especially when you are exercising

Drinking water helps your muscle cells work better

- Carry a water bottle with you all day
- Make frequent trips to the water cooler



If you don't like to drink water try:

- Herb tea, Crystal Light™, unsweetened iced tea, clear soups, oranges
- Flavor water with lemon or orange slices

4. Alcohol

The brain relies on glucose (sugar) for energy and a lack of glucose can lead to permanent brain damage and death. Normally blood sugars are very tightly controlled by the body; however drinking alcohol can cause low blood sugar in people with low glucose stores. This is because the job of processing the alcohol takes priority over the usual job of making glucose.

If you do choose to have alcohol then limit yourself to **one serving** and have a carbohydrate containing snack along with it.

One serving of alcohol equals:

12 ounces of beer/1 bottle of beer, 5 ounces of wine or 1½ ounces of spirits, such as vodka or whisky.

Sick days

If you are sick and not eating very much you need to try and eat/drink high calorie foods/ high carbohydrate foods often. Try to have something to eat every 2 hours. You must not go longer than 2 hours without eating/drinking calorie containing foods or beverages.

Ideas of foods to eat when you are sick:

- Jell-O or Jell-O pudding
- Sorbet or sherbet
- Canned fruit or fresh fruit
- Non-cola soft drinks – 7 up, gingerale, Sprite etc
- Juices, Kool-Aid, lemonade, Tang, Vegetable juice, Gatorade
- Popsicles, frozen fruit ice bars
- Fruit leathers or Energy-to-Go bars
- Oven baked bannock /bread/toast/bagels/pita bread with jam
- Rice, plain potatoes or cous cous
- Cold cereal with milk
- Pasta/noodles
- Oatmeal/cream of wheat with skim milk
- Plain crackers (melba toast, soda crackers, saltines)
- Cereal bars
- A glass of skim milk (or make the skim milk into chocolate milk or hot chocolate by adding Nestle Chocolate Quik Powder)
- Sugar, syrup, jam, jelly, honey, marmalade, plain hard candies, gum drops, jelly beans, mints (not chocolate), marshmallows, cranberry sauce

What if I can't keep food down?

It is important to give your body fuel in the form of carbohydrate. Try to drink something **every 30 minutes to one hour.**

Try sips of diluted juice, ginger ale, broth, tea with sugar, pop, Gatorade, Jell-O, Popsicles, frozen juice cubes.

Once your stomach has settled a bit, nibble on solid food such as crackers, dry toast, canned fruit, crispy rice snacks/rice cakes, arrowroot cookies or plain pasta (or any of the foods listed above).

If you are still unable to keep foods/fluid down please call the Metabolic Clinic at 604-875-5965.

How can I adapt my diet to participate in ceremonies that traditionally include fasting and exercise?

Fasting is a common practice in traditional spiritual ceremonies; however it can be very dangerous for a person with the CPT 1A variant, diabetes, or both conditions. Ask the people leading the ceremony if it is OK to drink sugared water every hour to prevent serious health problems. This will help you to achieve your ceremonial purposes.

What are the long-term complications of having the CPT 1A variant?

We do not know if there are long term consequences of the CPT 1A variant in adults. It is possible that the CPT 1A variant may put adults at risk of problems with low blood sugars and it is also possible that it may be related to other blood sugar problems like diabetes. Further research is needed to understand more about the consequences of this variant.

Staying healthy

It is important to watch any changes to the body that may result from the CPT 1A variant. Some people with the CPT 1A variant may have regular appointments at the metabolic clinic. For other people with the CPT 1A variant, it may be enough simply to have the contact information for the metabolic clinic so that you can call and speak to the doctors and nurses at the clinic if you have questions. Talk to the metabolic physician about what follow-up is recommended for you. The registered nurse at the Adult Metabolic Clinic will give you a wallet card to let medical staff know what to do in case of emergency.

One of the most important ways to stay healthy is to exercise regularly. It is safe for people with CPT 1A to exercise.

How can friends and family help?

Family, friends and community members play a special role in supporting the person with the CPT 1A variant.

It is important for everyone close to you to learn to recognize the signs and symptoms of a low blood sugar episode and to know how to treat it.

References:

Sinclair, Graham (August 31, 2011) personal communication

Star-G: Screening, Technology and Research in Genetics, Carnitine palmitoyl transferase deficiency (CPT 1A)

<http://www.newbornscreening.info/Parents/fattyaciddisorders/CPT1.html>

Helpful resources

Aboriginal Patient Navigators

Aboriginal Patient Navigators help aboriginal patients through the health system and offer support in other social health determinant areas such as housing and income.

- When you are booking your appointment, please ask if you'd like us to arrange for a Patient Navigator to come with you to your next appointment

Tel: 604-875-5600 ext. 63292

Toll-free: 1-877-875-1131



The Adult Metabolic Diseases Clinic

Vancouver General Hospital

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Fax: 604-875-5967

The National Organization for Rare Disorders, Inc.

P.O. Box 8923

New Fairfield, CT, 06812-8923

Tel: 1-800-999-6673

Website: www.rarediseases.org/

Fatty Oxidation Disorders (FOD) Family Support Group

Website: <http://www.fodsupport.org>

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