CARBOHYDRATE METABOLISM TEST

XXXXXXXXXXXX

Date of birth: XXXXXXX

Code: XXXXXX

WHAT THE REPORT INCLUDES

- Detailed EXPLANATION of the test performed and recommendations to be followed.
- SUMMARY TABLE presenting the metabolic areas analysed and the results from the DNA analysis, providing a quick overview of an individual's overall health status and highlighting any potential issues.
- BIBLIOGRAPHY providing scientific references for the test.

COLOURS USED

It indicates that the variants identified in the analysis do not unfavourably alter enzymatic activity of the proteins they encode and/or the risk associated with certain diseases.

It indicates that the variants identified in the analysis slightly unfavourably alter enzyme activity and/or the risk associated with certain disorders or diseases.

It indicates that the variants identified in the analysis alter enzyme activity in a particularly unfavourable way, resulting in an increased risk of developing certain disorders or associated diseases.

The results shown, as well as the considerations and explanations contained in the following pages of this booklet, should not be regarded as a medical diagnosis. It is important to bear in mind that genetic information is only a part of the total information needed to gain a complete picture of a person's state of health, and the data reported here is therefore a tool available to the treating physician to formulate a correct assessment of the patient's physiological state and suggest an appropriate personalised treatment.

INTRODUCTION

Carbohydrate metabolism is a multifaceted process that involves several genes and biochemical pathways responsible for the digestion, absorption, and utilization of carbohydrates to produce energy. Genetic variations can significantly impact how the body processes these nutrients, influencing susceptibility to conditions like insulin resistance, glucose intolerance, and the risk of developing type 2 diabetes.

This genetic test examines specific variants in genes related to carbohydrate metabolism, offering personalized insights into how the body handles sugar and starch intake. Understanding your genetic predisposition can guide you in adopting tailored dietary and lifestyle strategies to optimize metabolic health and reduce the risk of blood sugar imbalances.

It's important to note that the results should be considered alongside other factors, such as diet, lifestyle, and family history. While this information is not a medical diagnosis, it serves as a valuable tool to help personalize your nutritional and lifestyle choices, with the goal of improving overall metabolic health.

Testing for carbohydrate metabolism

Genetic analysis of the PPARG, KCNJ11, and TCF7L2 genes focuses on evaluating an individual's predisposition to carbohydrate metabolism and the risk of developing metabolic disorders, such as insulin resistance and type 2 diabetes.

- **PPARG (Peroxisome Proliferator-Activated Receptor Gamma)**: This gene regulates lipid and carbohydrate metabolism, influencing insulin sensitivity. Certain genetic variants are linked to an increased risk of insulin resistance and obesity.
- **KCNJ11 (Potassium Inwardly Rectifying Channel Subfamily J Member 11)**: This gene encodes a protein that helps regulate insulin secretion from pancreatic beta cells. Mutations in KCNJ11 may affect glycaemic responses and contribute to the risk of diabetes.
- **TCF7L2 (Transcription Factor 7 Like 2)**: This gene plays a pivotal role in regulating insulin secretion and glucose sensitivity. Some genetic variants in TCF7L2 are among the strongest known genetic risk factors for type 2 diabetes.

By analysing these genes, it becomes possible to identify genetic predispositions and adopt personalized strategies to improve carbohydrate metabolism through diet and lifestyle changes.

YOUR RESULT:

Lab ID	Gene	Allelic variants	Genotype		Result
Carbohydrate metabolism					
GTS009	PPARG	G	G	G	FAVOURABLE
	(Peroxisome Proliferator-Activated Receptor Gamma)	C			
WHAT YOUR GENETICS SAY					
					
A FAVOURABLE genetic profile is present for the gene analysed.					

Gentras ID	Gene	Allelic variants	Genotype		Result
Carbohydrate metabolism					
GTS010	KCNJ11	C	C	C	FAVOURABLE
(Potassium Inwardly Rectifying Channel Subfamily J Member		T			

WHAT YOUR GENETICS SAY

A FAVOURABLE genetic profile is present for the gene analysed.

Gentras ID	Gene	Allelic variants	Genotype		Result
Carbohydrate metabolism					
GTS040	TCFL7L	C	C	C	FAVOURABLE
(Transcription Factor 7 Like 2)		T			

WHAT YOUR GENETICS SAY

A FAVOURABLE genetic profile is present for the gene analysed.

EFFECTS OF UNFAVOURABLE VARIANTS for the PPARG, KCNJ11 and TCF7L2 genes

Unfavourable genetic variants in the PPARG, KCNJ11, and TCF7L2 genes can negatively affect carbohydrate metabolism, increasing the risk of developing metabolic disorders. The effects of these variants on carbohydrate metabolism and their association with conditions like insulin resistance and type 2 diabetes are as follows:

- **PPARG (Peroxisome Proliferator-Activated Receptor Gamma):**
  - Decreased insulin sensitivity, heightening the risk of insulin resistance.
  - Greater tendency to accumulate body fat, particularly in the abdominal region.
  - Elevated risk of obesity and type 2 diabetes, especially when combined with a diet high in saturated fats.
  - Reduced effectiveness of PPARG agonist medications (e.g., thiazolidinediones used for diabetes treatment).

- **KCNJ11 (Potassium Inwardly Rectifying Channel Subfamily J Member 11):**
  - Altered potassium channel function in pancreatic beta cells, leading to impaired insulin secretion.
  - Reduced pancreatic ability to adjust to fluctuations in blood glucose levels, raising the risk of hyperglycaemia.
  - Increased susceptibility to type 2 diabetes, particularly among individuals with a higher BMI.
  - Potential reduction in the effectiveness of drugs that target potassium channels, such as sulfonylureas (common anti-diabetic medications).

- **TCF7L2 (Transcription Factor 7 Like 2):**
    - Disrupted regulation of insulin secretion from pancreatic beta cells.
    - Increased risk of insulin resistance and reduced capacity to manage blood glucose.
    - Higher risk of developing type 2 diabetes, regardless of factors such as body weight.
    - Impaired insulin response following carbohydrate consumption, leading to higher glucose levels post-meal.
    - Decreased efficacy of certain medications like GLP-1 agonists, which stimulate insulin secretion.

In conclusion, these genetic variants can predispose individuals to disorders related to carbohydrate metabolism. However, their impact is influenced by environmental factors like diet and physical activity. Understanding these genetic predispositions enables the development of personalized strategies to prevent or manage insulin resistance and type 2 diabetes.

RECOMMENDED SOLUTIONS:

To counteract the effects of unfavourable variants in the PPARG, KCNJ11, and TCF7L2 genes, the following dietary and lifestyle changes, as well as therapeutic options, are recommended:

PPARG (Peroxisome Proliferator-Activated Receptor Gamma)

Goal: Improve insulin sensitivity and reduce fat accumulation.

1. **Dietary Recommendations:**
    - Limit saturated fats (e.g., red meat, whole milk, fried foods), which can worsen insulin resistance.
    - Increase unsaturated fats (e.g., olive oil, oily fish, nuts) to support PPARG function.
    - Follow a low glycaemic index diet with whole grains, vegetables, and lean proteins to stabilize blood glucose.
    - Consume more soluble fibre (e.g., oats, legumes, flaxseeds) to aid in carbohydrate absorption.
2. **Lifestyle Recommendations:**
    - Engage in regular physical activity, with a focus on resistance training (e.g., weightlifting) and aerobic exercise (e.g., brisk walking, swimming) to enhance insulin sensitivity.
    - Monitor body weight, as excess weight can amplify the effects of the unfavourable variant.
3. **Potential Therapeutic Strategies:**
    - In cases of diabetes or insulin resistance, healthcare providers may consider insulin-sensitizing medications such as metformin or thiazolidinediones.

KCNJ11 (Potassium Inwardly Rectifying Channel Subfamily J Member 11)

Goal: Support pancreatic function and enhance insulin secretion regulation.

1. **Dietary Recommendations:**
    - Distribute carbohydrate intake throughout the day in smaller, more frequent meals to avoid glycaemic spikes.
    - Avoid simple sugars (e.g., sweets, sugary drinks) that can overload the pancreas.
    - Increase intake of magnesium and potassium (e.g., dried fruits, leafy greens, bananas) to support the ion channels involved in insulin secretion.
2. **Lifestyle Recommendations:**

- Engage in regular physical activity, particularly exercises that improve insulin sensitivity (e.g., high-intensity aerobic training).
- Manage stress levels, as chronic high cortisol can negatively affect pancreatic function.

3. **Potential Therapeutic Strategies:**
   - In cases of diabetes, a healthcare provider may recommend medications that enhance insulin secretion, such as sulfonylureas or DPP-4 inhibitors.

TCF7L2 (Transcription Factor 7 Like 2)

Goal: Improve insulin secretion and optimize blood glucose response.

1. **Dietary Recommendations:**
   - Follow a low glycaemic load diet, balancing complex carbohydrates, proteins, and healthy fats to avoid blood sugar surges.
   - Avoid refined carbohydrates (e.g., white bread, sweets) that may worsen impaired insulin secretion.
   - Increase consumption of polyphenols and antioxidants (e.g., green tea, berries, bitter cocoa) to protect pancreatic cells from oxidative damage.
   - Include legumes and high-fibre foods to slow glucose absorption.
2. **Lifestyle Recommendations:**
   - Incorporate regular physical activity, which helps compensate for reduced insulin secretion.
   - Maintain a healthy body weight to improve overall glycaemic management.
3. **Potential Therapeutic Strategies:**
   - In cases of diabetes, doctors may consider GLP-1 agonists or DPP-4 inhibitors to better stimulate insulin secretion.
   - Regular blood glucose monitoring is recommended, even if symptoms are not evident.

In summary, if one has unfavourable variants of these genes, adopting a healthy lifestyle and a targeted diet can significantly reduce the risk of developing metabolic disorders. The personalised approach, based on genetic results, can optimise carbohydrate metabolism, and improve long-term health.

Carbohydrate sources:

Carbohydrates are primarily found in the following food groups:

- **Cereals and Grains:** Bread, pasta, rice, spelt, barley, oats, millet, quinoa, rye, maize, polenta, couscous, crackers, and cereal flakes.
- **Legumes:** Beans, lentils, chickpeas, peas, broad beans, soy, and lupins.
- **Tubers and Roots:** Potatoes, cassava, yam, Jerusalem artichoke, and yuca.
- **Fruits:** Bananas, apples, pears, grapes, dates, figs, mangoes, pineapples, cherries, apricots, peaches, kiwis, oranges, tangerines, strawberries, blueberries, and raspberries.
- **Vegetables:** Carrots, beets, squash, onions, tomatoes, and peppers.
- **Dairy Products:** Milk, yogurt, and fresh cheeses such as cottage cheese, which contain lactose.
- **Sweets and Processed Products:** White and brown sugar, honey, syrups, jams, cakes, biscuits, snacks, sugary cereals, milk chocolate, ice cream, and candy. Additionally, sugary drinks like fruit juices, soft drinks, sweetened tea, coffee, and energy drinks are rich in simple carbohydrates.

Carefully managing carbohydrate intake from these sources can play a key role in optimizing carbohydrate metabolism and reducing the risk of metabolic disorders.

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