Geneticheck - Genetic Report

Gene

HNF1A

Associated Diseases

Diabetes Mellitus, Noninsulin-dependent
Diabetes Mellitus, Insulin-dependent, 20
Diabetes Mellitus, Insulin-dependent-1
Hyperinsulinism Due To Hnf1a Deficiency
Mody
Renal Cell Carcinoma, Nonpapillary
Hepatic Adenomas, Familial
Maturity-onset Diabetes Of The Young, Type Iii

Phenotype

Feeding difficulties
Impaired ability to eat related to problems gathering food and getting ready to suck, chew, or swallow it.

Hypoglycemic seizures

Autoimmunity
The occurrence of an immune reaction against the organism's own cells or tissues.

Decreased circulating free fatty acid level

Glycosuria
An increased concentration of glucose in the urine.

Elevated hemoglobin A1c
An increased concentration of hemoglobin A1c (HbA1c), which is the product of nonenzymatic attachment of a hexose molecule to the N-terminal amino acid of the hemoglobin molecule. This reaction is dependent on blood glucose concentration, and therefore reflects the mean glucose concentration over the previous 8 to 12 weeks. The HbA1c level provides a better indication of long-term glycemic control than one-time blood or urinary glucose measurements.

Polyuria
An increased rate of urine production.

Episodic hyperhidrosis
Intermittent episodes of abnormally increased perspiration.

Neurodevelopmental abnormality
A deviation from normal of the neurological development of a child, which may include any or all of the aspects of the development of personal, social, gross or fine motor, and cognitive abilities.

Neonatal hypotonia
Muscular hypotonia (abnormally low muscle tone) manifesting in the neonatal period.

Exocrine pancreatic insufficiency
Impaired function of the exocrine pancreas associated with a reduced ability to digest foods because of lack of digestive enzymes.

Tachycardia
A rapid heartrate that exceeds the range of the normal resting heartrate for age.

Ketoacidosis
Acidosis resulting from accumulation of ketone bodies.

Hyperinsulinemic hypoglycemia
An increased concentration of insulin combined with a decreased concentration of glucose in the blood.

Loss of consciousness

Autosomal dominant inheritance
A mode of inheritance that is observed for traits related to a gene encoded on one of the autosomes (i.e., the human chromosomes 1-22) in which a trait manifests in heterozygotes. In the context of medical genetics, an autosomal dominant disorder is caused when a single copy of the mutant allele is present. Males and females are affected equally, and can both transmit the disorder with a risk of 50% for each child of inheriting the mutant allele.

Fasting hypoglycemia

Agitation
A state of exceeding restlessness and excessive motor activity associated with mental distress or a feeling of inner tension.

Maturity-onset diabetes of the young
The term Maturity-onset diabetes of the young (MODY) was initially used for patients diagnosed with fasting hyperglycemia that could be treated without insulin for more than two years, where the initial diagnosis was made at a young age (under 25 years). Thus, MODY combines characteristics of type 1 diabetes (young age at diagnosis) and type 2 diabetes (less insulin dependence than type 1 diabetes). The term MODY is now most often used to refer to a group of monogenic diseases with these characteristics. Here, the term is used to describe hyperglycemia diagnosed at a young age with no or minor insulin dependency, no evidence of insulin resistence, and lack of evidence of autoimmune destruction of the beta cells.

Drowsiness
Excessive daytime sleepiness.

Abnormality of the genitourinary system
The presence of any abnormality of the genitourinary system.

Maternal diabetes

Maternal diabetes can either be a gestational, mostly type 2 diabetes, or a type 1 diabetes. Essential is the resulting maternal hyperglycemia as a non-specific teratogen, imposing the same risk of congenital malformations to pregnant women with both type 1 and type2 diabetes.

Obesity
Accumulation of substantial excess body fat.

Hypoinsulinemia
A decreased concentration of insulin in the blood.

Pancreatic hypoplasia
Hypoplasia of the pancreas.

Abnormal insulin level
An abnormal concentration of insulin in the body.

Syncope
Syncope refers to a generalized weakness of muscles with loss of postural tone, inability to stand upright, and loss of consciousness. Once the patient is in a horizontal position, blood flow to the brain is no longer hindered by gravitation and consciousness is regained. Unconsciousness usually lasts for seconds to minutes. Headache and drowsiness (which usually follow seizures) do not follow a syncopal attack. Syncope results from a sudden impairment of brain metabolism usually due to a reduction in cerebral blood flow.

Hypoglycemic coma

Glucose intolerance
Glucose intolerance (GI) can be defined as dysglycemia that comprises both prediabetes and diabetes. It includes the conditions of impaired fasting glucose (IFG) and impaired glucose tolerance (IGT) and diabetes mellitus (DM).

Large for gestational age
The term large for gestational age applies to babies whose birth weight lies above the 90th percentile for that gestational age.

Palpitations
A sensation that the heart is pounding or racing, which is a non-specific sign but may be a manifestation of arrhythmia.

Pallor
Abnormally pale skin.

Decreased level of 1,5 anhydroglucitol in serum
A decrease in the level of 1,5 anhydroglucitol in the serum. 1,5-Anhydrosorbitol is a validated marker of short-term glycemic control. This substance is derived mainly from food, is well absorbed in the intestine, and is distributed to all organs and tissues.

Polycystic ovaries

Type II diabetes mellitus
A type of diabetes mellitus initially characterized by insulin resistance and hyperinsulinemia and subsequently by glucose interolerance and hyperglycemia.

Hyperinsulinemia
An increased concentration of insulin in the blood.

Hepatocellular adenoma
A benign tumor of the liver of presumably epithelial origin.

Lethargy
A state of disinterestedness, listlessness, and indifference, resulting in difficulty performing simple tasks or concentrating.

Abnormality of the kidney
An abnormality of the kidney.

Renal cyst
A fluid filled sac in the kidney.

Transient neonatal diabetes mellitus

Hypoketotic hypoglycemia
A decreased concentration of glucose in the blood associated with a reduced concentration of ketone bodies.

Intrauterine growth retardation
An abnormal restriction of fetal growth with fetal weight below the tenth percentile for gestational age.

Reactive hypoglycemia
Hypoglycermia following a meal (or more generally, after intake of glucose).

Excessive insulin response to glucagon test
An abnormally high increase in insulin levels following a glucagon stimulation test.

Abnormal oral glucose tolerance
An abnormal resistance to glucose, i.e., a reduction in the ability to maintain glucose levels in the blood stream within normal limits following oral administration of glucose.

Hepatomegaly
Abnormally increased size of the liver.

Overweight
Increased body weight with a body mass index of 25-29.9 kg per square meter.

Small for gestational age
Smaller than normal size according to sex and gestational age related norms, defined as a weight below the 10th percentile for the gestational age.

Neonatal hypoglycemia

Type I diabetes mellitus
A chronic condition in which the pancreas produces little or no insulin. Type I diabetes mellitus is manifested by the sudden onset of severe hyperglycemia with rapid progression to diabetic ketoacidosis unless treated with insulin.

Hyperglycemia
An increased concentration of glucose in the blood.

Increased C-peptide level
An elevated concentration of C-peptide in the circulation. Since C-peptide is secreted in equimolar amounts to insulin, this feature correlates with increased insulin secretion.

Nephropathy
A nonspecific term referring to disease or damage of the kidneys.

Retinopathy
Any noninflammatory disease of the retina. This nonspecific term is retained here because of its wide use in the literature, but if possible new annotations should indicate the precise type of retinal abnormality.

Insulin-resistant diabetes mellitus
A type of diabetes mellitus related not to lack of insulin but rather to lack of response to insulin on the part of the target tissues of insulin such as muscle, fat, and liver cells. This type of diabetes is typically associated with increases both in blood glucose concentrations as will as in fasting and postprandial serum insulin levels.

Heterogeneous

Sporadic
Cases of the disease in question occur without a previous family history, i.e., as isolated cases without being transmitted from a parent and without other siblings being affected.

Polydipsia
Excessive thirst manifested by excessive fluid intake.

Polyphagia
A neurological anomaly with gross overeating associated with an abnormally strong desire or need to eat.

Renal cell carcinoma
A type of carcinoma of the kidney with origin in the epithelium of the proximal convoluted renal tubule.

Diabetes mellitus
A group of abnormalities characterized by hyperglycemia and glucose intolerance.

Increased waist to hip ratio
Increased waist-to-hip ratio (WHR) is a measurement above the average for the dimensionless ratio of the circumference of the waist to that of the hips. WHR is calculated as waist measurement divided by hip

measurement.

Ketotic hypoglycemia
Low blood glucose is accompanied by elevated levels of ketone bodies in the body.

Insulin resistance
Increased resistance towards insulin, that is, diminished effectiveness of insulin in reducing blood glucose levels.

Abnormal C-peptide level
An anomolous circulating concentration of the connecting (C) peptide, which links the insulin A and B chains in proinsulin, providing thereby a means to promote their efficient folding and assembly in the endoplasmic reticulum during insulin biosynthesis. After cleavage of proinsulin, C-peptide is stored with insulin in the soluble phase of the secretory granules and is subsequently released in equimolar amounts with insulin, providing a useful independent indicator of insulin secretion.