Swiss Center for Genetics
RCLIN Swiss SA
Rue du Lac 10
1815 Clarens, Switzerland
+41 21 963 2500
genetics@rclin.com

SWISS CENTER FOR GENETICS

Patient:
First-name NAME:
DOB:
Gender: F/M

Analysis requested
By: Dr
Your reference:

Material:
Sample received:
Reporting date:
Lab reference:

GENETIC REPORT OF WOMAN'S HORMONAL HEALTH - SAMPLE

The Woman's Hormonal Health genetic testing investigates risks of hormonal metabolism disorders caused by certain gene polymorphisms. This test evaluates: sensitivity of estrogen receptors, synthesis of sex hormones, balance of active forms of sex hormones, metabolism of sex hormones (all three phases of detoxification), metabolism of hypothalamic-pituitary-adrenal system hormones.

Estrogen Metabolism - Phase I Detoxification — 94% activity

CYP1A1 | CYP1A1* | CYP1B1
CYP3A4 | CYP19A1 | CYP19A1*
CYP17A1

Metabolic Adaptation — 25% activity

ADRB2

Estrogen Metabolism - Phase II Detoxification — 60% activity

COMT | COMT* | MTHFR
GSTM1 | GSTP1 | GSTP1*
UGT1A1 | UGT1A6 | SULT1A1
SULT1E1 | SULT2A1 | NQO1

Nervous System — 63% activity

Energy
COMT | COMT* | MTHFR
MAOA | MAOB
Stress Resistance
PNMT
Dopamine Capacity
DRD2
Sleep/Wake Cycle Regulation
ADRB1

Estrogen Metabolism - Phase III Detoxification — 20% activity

ABCB1

Synthesis of sex hormones — 80% activity

Estrogen Synthesis
CYP19A1 | CYP19A1* | CYP17A1
Conversion of Testosterone to dihydrotestosterone
SRD5A2
Hormones Production
Testosterone / Estrogen / DHEA
CYP17A1

Sensitivity of Estrogen Receptors — 20% activity

ESR1 | ESR1* | ESR2

Color Key
Beneficial properties
Moderate risk
High risk
*2 Rs of the same gene

Brief Explanation of the Functions of the Studied Genes

Sensitivity of estrogen receptors

Mutations in a gene encoding the estradiol receptor may lead to increased sensitivity of cells to estradiol with its normal amount in the blood, and as a consequence, receptor relative hypoestrogenemia.

This has been shown to affect reproductive function, lead to disruption of calcium metabolism and disorganization of connective tissue.

Synthesis of sex hormones

In this section, we analyze the genes encoding the enzymes 17-alpha-hydroxylase and aromatase, which belong to the class of cytochromes. 17-alpha-hydroxylase is involved in the synthesis of all sex hormones (androgens and estrogens). Mutations in this gene can lead to increased hormone synthesis, i.e. hyperandrogenism and hyperestrogenism. Aromatase converts testosterone into estradiol (all female sex hormones are synthesized from male hormones). If aromatase activity is enhanced, this leads to increased synthesis of estradiol and hyperestrogenism.

Hyperestrogenism is a condition characterized by an excess of estrogens in the blood. Estrogens play the role of catalyst for the proliferation of the endometrium and mammary gland cells. Not only the reproductive system depends on them, but also the brain, bones and hair follicles. Hyperestrogenism can lead to such consequences as irregular or absent menstruation, insulin resistance, miscarriage, infertility, hypertension and atherosclerosis before the age of 50, osteoporosis and depression.

Hyperandrogenism is a syndrome caused by an increased amount of androgens in the peripheral blood and/or their increased activity. Among the symptoms of this disease in women are disruption of proper functioning of hair follicles and sebaceous glands (acne, seborrheic dermatitis, hirsutism), reproductive problems (algo menorrhea, oligomenorrhea, polymenorrhea, amenorrhea, infertility).

Deviations from the normal range of active forms of androgens/estrogens can cause many pathological conditions. These include an increased amount of dihydrotestosterone, one of the most active androgens, formed from testosterone directly in the "target organs" with the participation of a special enzyme (5-alpha-reductase). By controlling the activity of SRD5A2, such misbalances can be prevented.

Metabolism of sex hormones (3 phases of detoxification), balance of active forms of sex hormones

Sex hormones play a vital role in woman's health. It is important to note that despite the division of sex hormones into female and male, in the female body, androgens (male sex hormones) serve as precursors of estrogens (female sex hormones). Therefore, a deficiency of androgens in the female body is as undesirable as an excess thereof (hyperandrogenism).

The endometrium contains a very large number of estrogen receptors. High levels of estrogen receptor expression is a risk factor for development of endometriosis and endometrial neoplasms. An important target for treatment and prevention of endometrial diseases (endometriosis, malignant neoplasms) is aromatase, a key enzyme in the biosynthesis of estrogens, responsible for their biosynthesis from male sex hormones. On the other hand, metabolic activation with the formation of estrogen derivatives, sometimes having even greater proliferative activity compared to estrogens themselves, plays no less a role in endometrial dysplastic processes.

It has been established that androgens also affect the proliferation of endometrial cells. Dysregulation of androgen biosynthesis is associated with endometrial pathologies and dysfunction and plays a role in the development of infertility and miscarriages. Androgen receptors are one of the most important therapeutic targets for the prevention and treatment of endometrial disorders.

Estrogen metabolites can have a significant impact on the body; they are most often associated with the development of cancer, as they have a carcinogenic effect and damage cellular DNA.

The CYP1A1 gene is responsible for the formation of an enzyme that is involved in the metabolism of estrogens along a non-oncogenic pathway. Variants of the CYP1A1 gene can affect the rate and activity of estrogen metabolism and, therefore, an individual's predisposition to the development of cancer and other diseases.

The CYP1B1 gene encodes an enzyme that leads estrogens along a more dangerous detoxification pathway, where metabolites that can cause DNA damage to accumulate. They also increase oxidation and inflammation, which explains their association with PMS, oncology, menstrual cramps, headaches, and other pathologies.

The COMT gene encodes the enzyme catechol-O-methyltransferase, which is involved in the metabolism of dopamine, norepinephrine, and epinephrine. Variations in this gene can affect our response to stress, emotional states, and cognitive functions. COMT also participates in the inactivation of estrogens, transforming them into forms that are safe for the body. Mutations in this gene and decreased COMT activity are associated with increased risks of breast cancer, ovarian cancer, and other pathologies.

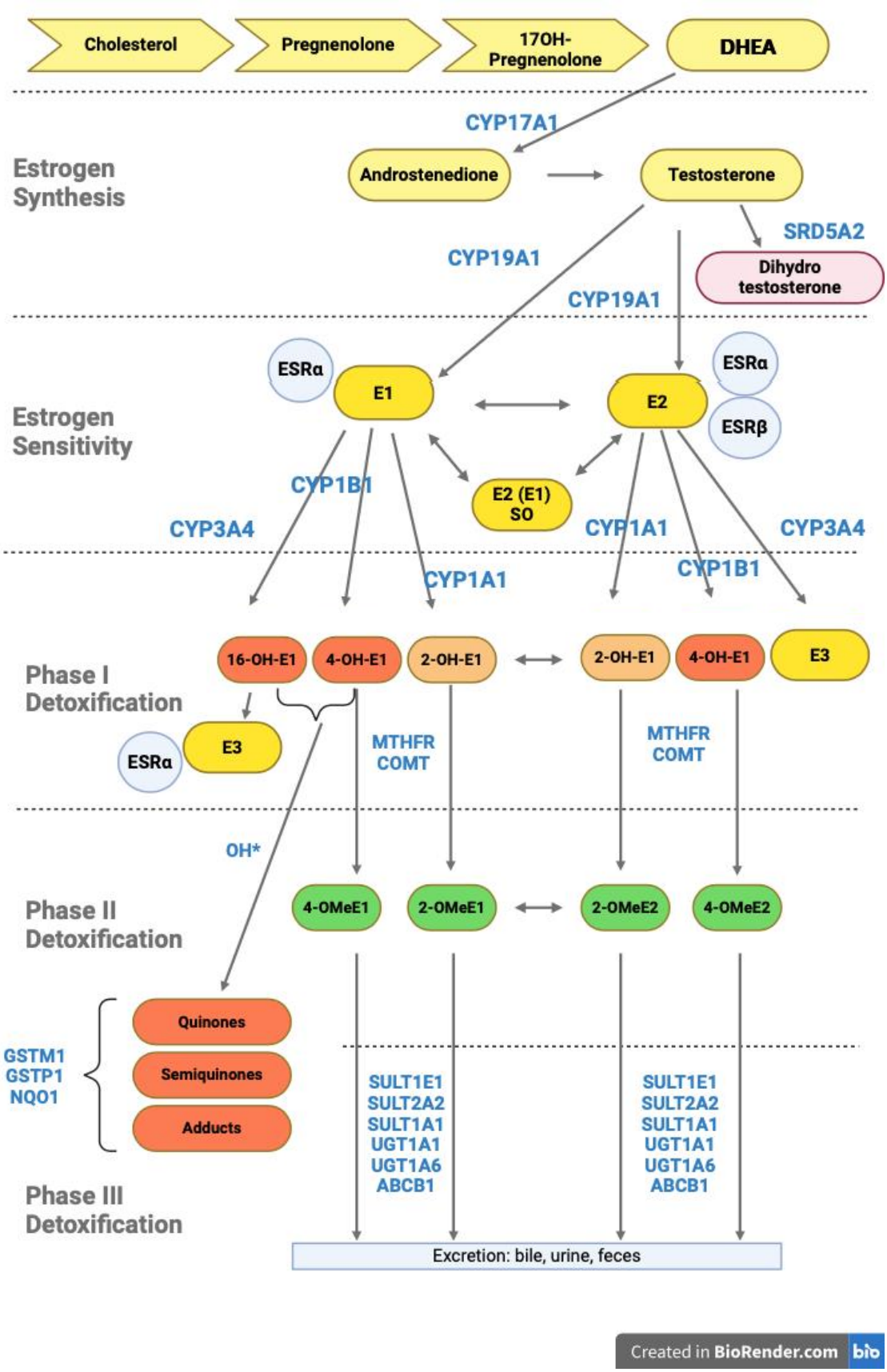
The function of the UGT1A1 gene is to convert estrogens into their conjugates with glucuronic acid. The conjugation process allows the body to safely metabolize and remove estrogens from the body. Some variants of the UGT1A1 gene can affect its activity and effectiveness. This can lead to the accumulation of estrogens in the body and increase the risk of developing certain diseases, such as breast cancer.

The function of the GSTT1 and GSTM1 genes is to convert estrogens into their conjugates with glutathione. Polymorphisms in the GSTT1 gene lead to a lack of enzyme activity, which, when combined with COMT deficiency, increases the concentrations of carcinogenic estrogen metabolites and increase the risk of hypoestrogenemia, reproductive disorders, and estrogen-dependent proliferative diseases.

Hypothalamic-pituitary-adrenal system hormone metabolism

The HPA axis is a major neuroendocrine system. It is responsible for regulating response to stress and other body processes, including digestion, immune responses, emotional regulation, sexual activity, as well as energy storage and expenditure.

Sex Steroid Hormones Metabolism



GENETIC ANALYSIS RESULTS of Sensitivity of estrogen receptors

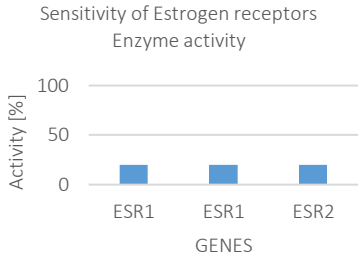
Role: Steroid hormones are a group of hormones that are synthesized from cholesterol.
Sensitivity of steroid receptors, such as estrogen, is associated with polymorphisms in the ESR1, ESR2 genes.
These genes encode receptors that interact with hormones and can affect various physiological processes.
Key Enzymes: ESR1: This polymorphism can affect the expression of the estrogen receptor and is associated with the risk of developing of various diseases, such as breast cancer and osteoporosis. ESR1 is associated with response to stress and estrogen production, as well as increased risk of estrogen-linked conditions.

ESR1: This polymorphism can also affect the function of the estrogen receptor and is associated with the risk of cardiovascular diseases, cancer, dyslipidaemia, hypertension, obesity, and cognitive brain function.
ESR2: Polymorphism in the ESR2 gene can affect sensitivity to estrogen and is associated with the risk of cancer and other diseases. ESR2 is activated by estrogen, with generally anti-cancer effects. This genotype has an increased risk of cardiovascular and metabolic conditions after menopause.

Genotypes and acticity of Sensitivity of estrogen receptors

GENES	Genomic Coordinates	Genotype	Zygosity	Enzyme activity
ESR1	Chr6: 151842246	GG	Homozygous	20
ESR1	Chr6:151842200	CC	Homozygous	20
ESR2	Chr14:64233098	TT	Homozygous	20

Global activity: 20%



GENETIC ANALYSIS RESULTS of Synthesis of sex hormones

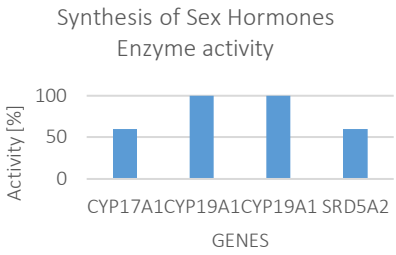
Role: Activity of enzymes influences the level of cortisol, androgens and estrogen.
Key Enzymes: CYP17A1 enzymatic activity shows conversion from progesterone to cortisol, androgens and estrogens. This activity can be up-regulated by stress, blood sugar dysregulation, excess insulin, and alcohol. Adequate physical activity and stress management needs to be ensured.

CYP19A1 enzymatic activity shows conversion of androgens into estrogens. Inflammation, high level of insulin, chronic stress will increase CYP19A1 activity.
SRD5A2 enzymatic activity shows conversion from testosterone to 5a-DHT. Controlling SRD5A2 activity gives benefit in cancer prevention. It is also beneficial for women with PCOS to protect against hair loss.

Genotypes and acticity of Synthesis of sex hormones

GENES	Genomic Coordinates	Genotype	Zygosity	Enzyme activity
CYP17A1	Chr10: 102837395	GT	Heterozygous	60
CYP19A1	Chr15: 51242350	TT	Homozygous	100
CYP19A1	Chr15: 51210789	GG	Homozygous	100
SRD5A2	Chr2: 31580636	CG	Heterozygous	60

Global activity: 80%



GENETIC ANALYSIS RESULTS of Phase I enzymes

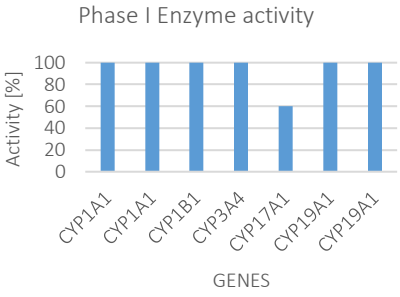
Role: Phase I detoxification involves the modification of toxins through chemical reactions, such as oxidation, reduction, and hydrolysis. These reactions introduce or expose a functional group (such as a hydroxyl group) on the toxin, making it more reactive.
Key Enzymes*:* Cytochrome P450 enzymes (CYPs):

These are the most important Phase I enzymes. They are a large family of enzymes that catalyze the oxidation of organic substances. CYP enzymes, such as CYP1A1, CYP1B1, CYP3A4, CYP17A1, and CYP19A1 play a major role in drug metabolism and detoxification of various toxins.

Genotypes and acticity of Phase I enzymes

GENES	Genomic Coordinates	Genotype	Zygosity	Enzyme activity
CYP1A1	Chr15: 74720644	TT	Homozygous	100
CYP1A1	Chr15: 74719300	AA	Homozygous	100
CYP1B1	Chr2: 38071060	CC	Homozygous	100
CYP3A4	Chr7:99784473	TT	Homozygous	100
CYP17A1	Chr10: 102837395	GT	Heterozygous	60
CYP19A1	Chr15: 51242350	TT	Homozygous	100
CYP19A1	Chr15: 51210789	GG	Homozygous	100

Global activity: 94%



GENETIC ANALYSIS RESULTS of Phase II enzymes

Role: Phase II of detoxification involves Sulphation, Conjugation with glutathione, Glucuronidation, Acetylation, Amino-Acid Conjugation, Methylation to form more stable and water-soluble compounds that can be easily excreted.

Key Enzymes: The UGT1A1 gene encodes UDP-glucuronyl transferase, the main enzyme responsible for the glucuronidation of bilirubin and some other compounds. The UGT1A1 gene is expressed mainly in the liver. Certain genetic variants affecting the production of UDP-glucuronyl transferase are a risk factor for the development of Gilbert's syndrome. The function of the UGT1A1 gene is to convert estrogens into their conjugates with glucuronic acid. The conjugation process allows the body to safely metabolize and eliminate estrogens from the body.

Glutathione-S-transferases (GSTs): Catalyze the conjugation of glutathione to reactive intermediates, neutralizing them and making them more water-soluble.

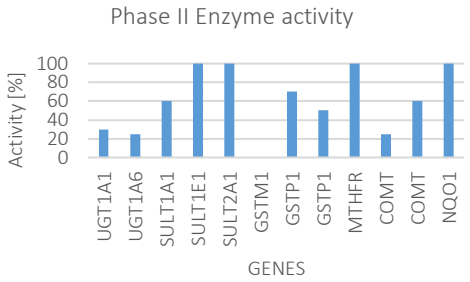
Glutathione S-transferase Pi (GSTP1) is an isozyme encoded by the GST pi gene that plays an important regulatory role in detoxification, anti-oxidative damage, and the occurrence of various diseases. Polymorphisms of this gene reduce the activity of the enzyme, which inhibits Phase II xenobiotics, including detoxification of medicinal derivatives.

Sulfotransferases (SULTs): Transfer sulphate groups to hydroxyl groups on toxins, increasing their water solubility. Sulfotransferase enzymes family (SULT1A1, SULT1E1, SULT2A1) catalyze the sulphate conjugation of many hormones, neurotransmitters, drugs, and xenobiotic compounds.

The end products of Phase II detoxification are generally non-toxic, highly water-soluble, and can be readily excreted from the body through urine or bile.

Genotypes and acticity of Phase II enzymes

GENES	Genomic Coordinates	Genotype	Zygosity	Enzyme activity
Glucoronidation				
UGT1A1	Chr2: 233764076	GG	Homozygous	30
UGT1A6	Chr2:233693631	TT	Homozygous	25
Sulphation				
SULT1A1	Chr16:28606193	TC	Heterozygous	60
SULT1E1	Chr4: 69860103	CC	Homozygous	100
SULT2A1	Chr19:47868938	TT	Homozygous	100
Glutathion Conjugation				
GSTM1	Null Allelle	AA	Homozygous	0
GSTP1	Chr11: 67585218	GA	Heterozygous	70
GSTP1	Chr11:67586108	TT	Heterozygous	50
Methylation				
MTHFR	Chr1: 11796321	GG	Homozygous	100
COMT	Chr22: 19963748	AA	Homozygous	25
COMT	Chr22: 19962712	CT	Heterozygous	60
NQO1	Chr16: 69711242	GG	Homozygous	100



Global activity: 60%

GENETIC ANALYSIS RESULTS of Phase III enzymes

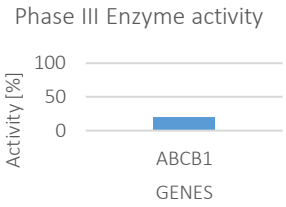
Role: Phase III detoxification involves elimination of water-soluble molecules out of the body through bile/intestine (stool), kidneys (urine) and skin (sweat).

Key Enzymes: ABCB1 enzyme regulates Phase III of the detoxification process and allows toxins to move across cellular barriers in the liver, gastrointestinal system, kidneys, and the blood-brain barrier.

The ABCB1 gene regulates the production of important ATP-dependent Phase 3 antiporter protein responsible for transporting various xenobiotics, drugs, lipids, and other exogenous and endogenous toxins out of cells for excretion. It can also be found under the name of MultiDrug Resistance 1 (MDR1) or the P-Glycoprotein (P-GP).

Genotypes and acticity of Phase III enzymes

GENES	Genomic Coordinates	Genotype	Zygosity	Enzyme activity
ABCB1	Chr7:87509329	GG	Homozygous	20



Global activity: 20%

GENETIC ANALYSIS RESULTS of Nervous System / Stress Resistance

Role: Neurotransmitter imbalance causes fatigue, mood swings, sleep disorders, depression and aggression. Neurotransmitter levels relate to adrenaline, noradrenaline, dopamine, serotonin whose levels depend on the methylation processes and hypothalamic-pituitary-adrenal system activity.

Key Enzymes: Variants on COMT, MAOA & MAOB, MTHFR relate to neurotransmitter levels and lead to imbalances causing mood swings and chronic fatigue.

Gene expression of PNMT (phenyl-ethanolamine N -methyl transferase), the enzyme catalyzing the transfer of a methyl group from S-adenosyl-L-methionine (SAM) to norepinephrine in the final step of the biosynthesis of epinephrine.

DRD2 gene is responsible for regulating synthesis, storage and It has been shown that mutation of DRD2 gene can increase the risk of developing addiction and neuropsychiatric diseases. Moreover, it can inhibit dopamine production and activity, leading to psychiatric and psychotic effects. The function of each dopamine receptor D2: attention, sleep, memory, learning.

The ADRB1 gene encodes for the β1-adrenergic receptor. These receptors respond to hormones (including the hormones that regulate the sleep cycle). The ADRB1 gene is related to adrenaline and noradrenaline.

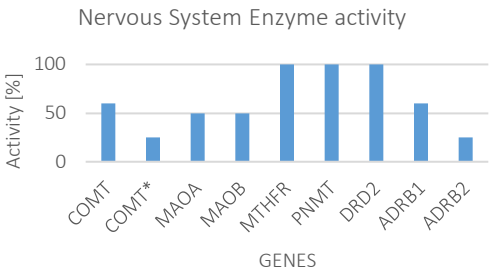
ADRB2 controls the physiological response to adrenaline, as it stimulates heart rate, blood flow, and availability of glucose for immediate energy.

release of dopamine.

Genotypes and acticity of Nervous System Enzymes

GENES	Genomic Coordinates	Genotype	Zygosity	Enzyme activity
Methylation/Energy				
COMT	Chr22: 19962712	CT	Heterozygous	60
COMT*	Chr22: 19963748	AA	Homozygous	25
MAOA	ChrX: 43731789	TT	Homozygous	50
MAOB	ChrX: 43768752	CC	Homozygous	50
MTHFR	Chr1: 11796321	GG	Homozygous	100
Methylation/Stress Resistance				
PNMT	Chr17:39668292	GG	Homozygous	100
HPA Axis/Dopamine Capacity				
DRD2	Chr11:113412966	CC	Homozygous	100
HPA Axis/Sleep-Wake Cycle Regulation				
ADRB1	Chr10:114045297	CG	Heterozygous	60
HPA Axis/Metabolic Adaptation				
ADRB2	Chr5: 148826877	GG	Homozygous	25

Global activity: 63%



Method

DNA was extracted from a cheek swab sample in accordance with the kit manufacturer's instructions. Genotyping was performed using Agena's MassARRAY © System. In a first step, raw results were analyzed using the MassARRAY Report software. In a second step, this report was generated using proprietary scripts.

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