Fagron AcneTest

Genetic awareness to personalize acne treatment

Fagron genomics

Patient Name	**Mariana Diaz Hernadez**
Date of Birth	**03-06-1974**
Gender	**Female**
Sample code	**NUT04947AA**
Sample date	**04-06-2020**
Date of the results	**04-06-2020**
Requesting physician	**Demo**
Requesting physician telephone	**Demo**

How to read and use the Fagron AcneTest report

This report is structured into the following sections:

I. Clinical questionnaire data

Here you will find the data entered in the clinical questionnaire for this patient.

II. Results Overview and Treatment

An artificial intelligence algorithm will generate a list of drugs indicated for acne treatment in that patient. Validated formulations will also be available here.

III. Detailed results

Results combining genetics and clinical aspects will be categorized into the following topics to improve the understanding of the acne presentation in the patient and guide treatment.

Results categories

- Skin Predisposition to acne
- Skin condition and inflammation
- Predisposition to hormone-related acne
- Nutritional correlation

IV. Complete genetic results

A list of the genotypes presented by the patient for each one of the 60 SNPs analyzed to fully understand the relevant genetic profile of that patient regarding acne.

V. Genetics and Acne

Here we explain basic concepts of the influence of genetics in the treatment of acne and its sequalae.

I.
Clinical Questionnaire Data

Data entered in the clinical questionnaire for this patient.

Patient personal information 1/2

Summary of clinical information acquired by the physician

PERSONAL DATA	
Age	38
Gender	Woman

BIOMETRIC DATA	
Weight (kg)	58
Height (cm)	166
BMI	23.72

MEDICAL HISTORY	
Systemic Hypertension	Yes/No
Diabetes Mellitus	Yes/No Controlled/Not-controlled
Dyslipidemia	Yes/No
Liver disease	Yes/no
Hormonal Diseases	Yes/No
Previous or current use of anabolic steroids	Yes/No Previous/Current
Humor disorders	Yes/No
Perfonal or familial of thromboembolic events	Yes/No
Cancer or neoplasia	Yes/No

GYNECOLOGICAL HISTORY	
Use of Contraception	Yes/No
Polycystic ovary syndrome	Yes/No
Gravida/Para/abordus	G_P_A_
Current pregnancy	Yes/no
Intention of pregnancy	Yes/No

SOCIAL HISTORY	
Exposure to sun and visible light	Yes/No
Physical activity	Yes/No Intense/not intense
Intake of refined carbohydrate	More than twice a day/Less than twice a day
Ethylism	Yes/no
Illicit drug use	Yes/no

Patient personal information 2/2

Summary of clinical information acquired by the physician

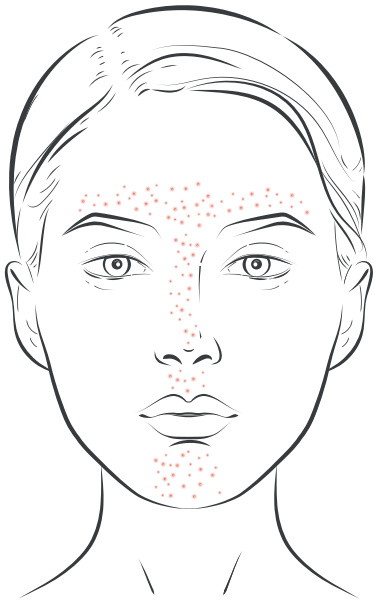
HISTORY OF PREVIOUS TREATMENTS	
Previous treatments	(here, we should have a list of the previous topic, oral and skin care treatemtns taken by the patient)
Previous skin procedures	(here we should have a list of the previous skin procedures with the date they were done)

LABORATORY EXAM RESULTS	
Laboratory results	
Creatinin	
Urea	
AST	
ALT	
Alcaline Phosphatase	
Gama-GT	
Bilirrubin	Total / Direct/ Indirect
Cholesterol	Total / LDL/ HDL
Tryglycerides	
CK	
Beta-HCG	
Fasting Glycemia	
Total testoterone	
Free Testosterone	
17-hydroxyprogesterone	
SDHEA	
SHBG	
Prolactine	
LH and FSH	LH / FSH

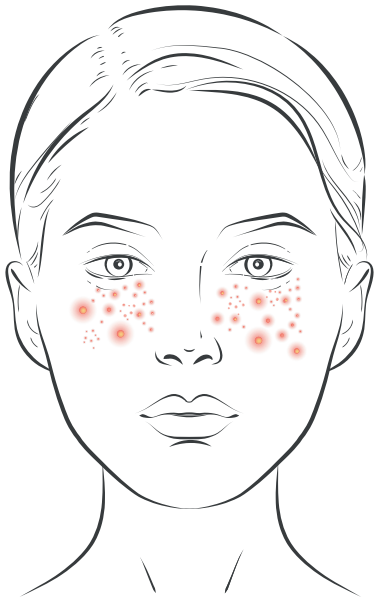
Patient acne classification

Description of the method

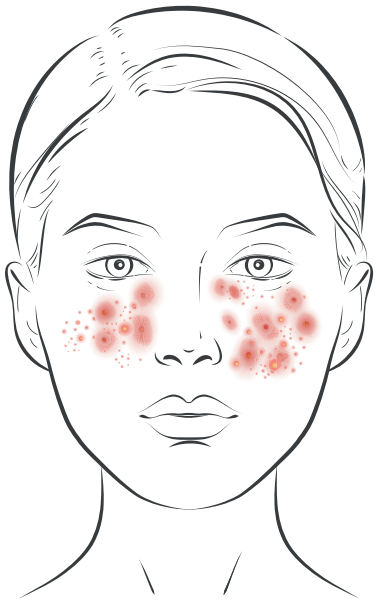
Acne classification



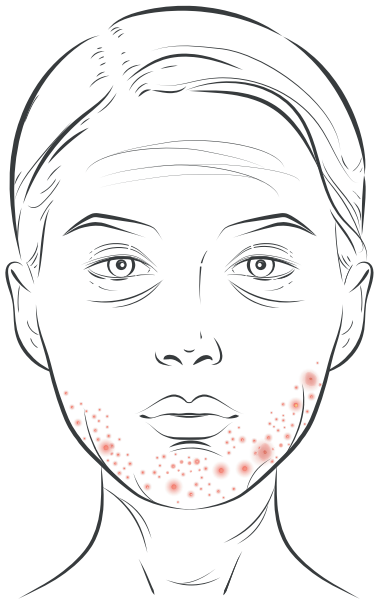
Grade I
(Comedogenic)



Grade II and III
(papular and pustular/inflammatory)



Grade IV
Grade IV (conglobata/nodulocystic)



Acne in an adult woman

II.
Results Overview and Treatment

List of drugs indicated for acne treatment and validated formulations.

Results Summary

Summary of the results generated by the genetic analysis

CATEGORY	DESCRIPTION	RESULT
Skin Predisposition to acne	Lorem ipsum dolor sit amet, consectetur adipiscing elit, sed do eiusmod tempor incididunt ut labore et dolore magna aliqua. Ut enim ad minim veniam, quis nostrud exercitation ullamco laboris nisi ut aliquip ex ea commodo consequat.	40.59%

• Predisposition to severe acne	MEDIUM RISK	Pg. 00

CATEGORY	DESCRIPTION	RESULT
Skin Condition and Inflammation	Lorem ipsum dolor sit amet, consectetur adipiscing elit, sed do eiusmod tempor incididunt ut labore et dolore magna aliqua. Ut enim ad minim veniam, quis nostrud exercitation ullamco laboris nisi ut aliquip ex ea commodo consequat.	76.86%

• Predisposition to scars and hyperpigmentation	MEDIUM RISK	Pg. 00
• Predisposition to increased sebum production	HIGH RISK	Pg. 00
• Predisposition to skin sensitivity	HIGH RISK	Pg. 00

CATEGORY	DESCRIPTION	RESULT
Predisposition to hormone-related acne	Lorem ipsum dolor sit amet, consectetur adipiscing elit, sed do eiusmod tempor incididunt ut labore et dolore magna aliqua. Ut enim ad minim veniam, quis nostrud exercitation ullamco laboris nisi ut aliquip ex ea commodo consequat.	63.17%

• Predisposition to acne due to hormonal alteration	MEDIUM RISK	Pg. 00
• Predisposition to polycystic ovary syndrome	HIGH RISK	Pg. 00

CATEGORY	DESCRIPTION	RESULT
Nutritional advice	Lorem ipsum dolor sit amet, consectetur adipiscing elit, sed do eiusmod tempor incididunt ut labore et dolore magna aliqua. Ut enim ad minim veniam, quis nostrud exercitation ullamco laboris nisi ut aliquip ex ea commodo consequat.	63.17%

• Predisposition to retinoid-related hyperlipidemia	MEDIUM RISK	Pg. 00
• Lipid metabolism	HIGH RISK	Pg. 00
• Carbohydrate metabolism	MEDIUM RISK	Pg. 00
• Food allergy	LOW RISK	Pg. 00

INDICATIONS

Low risk Medium risk High risk

Drug Efficacy Panel

Taking into consideration both the genetic predisposition and the clinical indication-specific for the patient, here are the most appropriate drugs to be used in the treatment. This panel of drugs is generated by the clinical and pharmacogenetic approach of the test, in which we employ the genetic information better understand how the patient responds to the treatment. They are here graded regarding the best suitability for this treatment.

Topical Antibiotics
• Clindamycin phosphate
• Erythromycin
• Azithromycin
• Benzoyl peroxide
• Metronidazole
• Dapsone
Retinoids
• Adapalene
• Tretinoin
• Isotretinoin
• Retinol
Antiandrogens
• Cyproterone acetate
• Spironolactone
• Flutamide
Alpha-adrenergic agonists
• Brimonidine
Depigmenting agents
• Azelaic acid
• Hydroquinone
• Alfa-arbutin
• Kojic acid
• Niacinamide
• Ellagic acid
• Tranexamic acid
• Sodium Ascorbyl phosphate
• Vitamin C
Parasiticides
• Ivermectin
• Permethrin
• Levamisole
Corticosteroids
• Clobetasol
Antifungals
• Miconazol
DCIs
Alpha hydroxyacids, Polyhydroxiacids, and other keratolytics
• Glycolic acid
• Mandelic acid
• Gluconolactone
• Kinetin (Adenin, N(6)-furfuriladenin)

Anti-inflammatory
• Alpha-bisabolol
• Phytosphingosine (and) Phytosphingosine HCl
• Aloe vera gel (200:1)
• Enoxolone (18-beta-glycyrhetinic acid)
• Green Tea (Camellia sinensis) aqueous or glycolic extracts
• EGCG (Epigallocatechin gallate)
Antiseptics
• Tea tree oil (Melaleuca alternifolia essential oil)
• Propolis glycolic or alcoholic extract
Sebolytics
• Zinc acetate
• Zinc pyrithione
Oral vitamins
• Vitamin A
• Vitamin C
• Vitamin D
• Vitamin E
• Vitamin B6
• Pantothenic acid (as calcium pantotenate)
Oral minerals
• Chromium (picolinate, chromium yeast)
• Copper (as gluconate, glycinate, sulfate)
• Selenium (as selenomethionine, selenium yeast)
• Zinc (as gluconate, sulfate)
Probiotics
• Bifidobacterium bifidum
• Lactobacillus acidophilus
• Lactobacillus bulgaricus
• Lactobacillus plantarum
• Lactobacillus rhamnosus
Phytoactives
• Guggul (Commiphora mukul) dry extract
• Silymarin extract (topical & systemic)
• Silibin® (Silymarin Phytosome) (systemic)
• Trans-resveratrol (topical)
• EGCG (Epigallocatechin gallate) (topical)
Fatty acids
• Gamma-linolenic acid (GLA) (Borage oil)
• Omega-3
Aminoacids and derivatics
• L-carnitine;
• N-acetylcysteine

INDICATIONS

The length of the green indicating from more to less recommended,
and those compounds we do not recommend from green to red, indicating less recommended.

Prescription Disclaimers

Antiandrogenic Treatment

The use of hormonal therapy might be related to the risk of thrombotic events. Caution should be applied when prescribing and following patients undergoing antiandrogenic therapy. Further clinical and laboratorial evaluation of the patient should be performed in order to mitigate that risk.

Antiandrogenic Treatment in Patients Undergoing Masculinizing Hormone Therapy

Currently, there are no guidelines directed to the transgender population, therefore, we must proceed with caution when beginning any antiandrogen treatments. It is important to not that the base of the acne treatment depends on the classification of the presented lesions, i.e. keratolytics for comedonian acne; fixed combinations and antibiotics (topical and oral) for inflammatory acne; and isotretinoin for severe cases. All medical decisions should be taken in accordance with the patient.

Formulations 1/2

A personalized formulation with suitable active ingredients and doses. The formulations below are composed of the validated combinations of molecules which are both active and safe to provide the best treatment possible for this patient.

Oral Treatment

Formula	
Astaxanthin	8 mg/day
Cystine	200 mg/day
Selenium yeast	100 mg/day
Oral	
Dosage 1 capsule per day, 90 capsules for 3 months.	

Formulations 2/2

A personalized formulation with suitable active ingredients and doses. The formulations below are composed of the validated combinations of molecules which are both active and safe to provide the best treatment possible for this patient.

Topical Treatment 1

Formula	
Latanoprost Fagron	0.005%
Tretinoin	0.01%
Dutasteride	0.25%
TrichoSol	100 ml

Dosage
Apply at night before bedtime. Leave the solution on your scalp for as long as possible.
Wash your scalp the next day.

Topical Treatment 2

Formula	
Latanoprost Fagron	0.005%
Tretinoin	0.01%
Dutasteride	0.25%
TrichoSol	100 ml

Dosage
Apply at night before bedtime. Leave the solution on your scalp for as long as possible.
Wash your scalp the next day.

Prescriptions 1/2

A personalized formulation with suitable active ingredients and doses. The formulations below are composed of the validated combinations of molecules which are both active and safe to provide the best treatment possible for this patient.

Oral Treatment

Formula	
Astaxanthin	8 mg/day
Cystine	200 mg/day
Selenium yeast	100 mg/day
Oral	
Dosage 1 capsule per day, 90 capsules for 3 months.	

Signature of the prescribing physician	
Dr	
Physician registtration No.	

Prescriptions 2/2

A personalized formulation with suitable active ingredients and doses. The formulations below are composed of the validated combinations of molecules which are both active and safe to provide the best treatment possible for this patient.

Topical Treatment 1

Formula	
Latanoprost Fagron	0.005%
Tretinoin	0.01%
Dutasteride	0.25%
TrichoSol	100 ml

Dosage
Apply at night before bedtime. Leave the solution on your scalp for as long as possible.
Wash your scalp the next day.

Signature of the prescribing physician	
Dr	
Physician registtration No.	

Topical Treatment 2

Formula	
Latanoprost Fagron	0.005%
Tretinoin	0.01%
Dutasteride	0.25%
TrichoSol	100 ml

Dosage
Apply at night before bedtime. Leave the solution on your scalp for as long as possible.
Wash your scalp the next day.

Signature of the prescribing physician	
Dr	
Physician registtration No.	

III. Detailed results

Combining genetics and clinical aspects, results will be categorized into the following topics to improve the understanding of the acne presentation in the patient and guide treatment.

1. Skin Predisposition

1.1 Predisposition to severe acne

– HIGH RISK –

ABOUT

As a disease with a significant inflammatory disease, polymorphisms in genes related to the immune response will significantly impact the acne presentation in a patient. The type and severity of lesions may be substantially influenced by genetics.

Acne grading as well as the presence of inflammatory lesions influence the appearance of long-lasting consequences, e.g., scars and post-inflammatory hyperpigmentation. Therefore, being predisposed to severe acne might be a determining factor to early initiate specific treatment.

CATEGORY	DESCRIPTION	RESULT
Predisposition to severe acne	Deficient metabolization of monounsaturated fatty acids (MUFA) intakes. Consumption of high levels of MUFA (>13% total calories) may result in an increased BMI.	**HIGH RISK**

HIGH RISK

This result indicates that the patient is predisposed to presenting severe acne.
The severity of lesions on the onset and genetic predisposition are essential determinants of sequelae, e.g., scars, hyperpigmentation, and relapse.
Therefore, appropriate treatment should be started early for this patient.

INDICATIONS

Low risk Medium risk High risk

2.
Skin condition and inflammation

2.1 Predisposition to scars and hyperpigmentation

– HIGH RISK –

ABOUT

As acne is tightly related to inflammation, genetic markers predisposing to more exacerbated inflammation are often associated with lesions' appearance and consequences.

The inflammatory immune environment is widely known to have consequences on the stimulation of both melanocytes and fibroblasts. Thus, the more inflammation happens during the acne process in a patient, the more this patient is likely to present scars and hyperpigmented spots.

CATEGORY	DESCRIPTION	RESULT
Predisposition to scars and hyperpigmentation	Genetic predisposition to exacerbated inflammation, resulting in being more prone to the formation of scars and hyperpigmentated areas	**HIGH RISK**

HIGH RISK

This result indicates the patient is at high risk for developing post-acne scars and hyperpigmented lesions. The recommendation of early treatment and lightning agents is usually helpful.

INDICATIONS

Low risk Medium risk High risk

2.
Skin condition and inflammation

2.2 Predisposition to increased sebum production

– HIGH RISK –

ABOUT

The production of sebum is one of the most widely known factors involved in the pathogenesis of acne. Although sebum is produced in response to several physical and chemical stimuli, there are genetic elements that might predict the predisposition to augmented sebum production. Thus, treatment might be planned accordingly.

CATEGORY	DESCRIPTION	RESULT
Predisposition to increased sebum production	Genetic predisposition to increased activity and secretion of the sebaceous glands	**HIGH RISK**

HIGH RISK

This result indicates this patient is at high risk of hyper seborrhea. It predisposes to the appearance of acne. However, the influence of this predisposition might be mitigated with the use of appropriate treatment, e.g., keratolytic agents.

INDICATIONS

Low risk Medium risk High risk

2.
Skin condition and inflammation

2.3 Predisposition to skin sensitivity

– HIGH RISK –

ABOUT

The treatment to the various presentations of acne might severely impact the skin condition leading to sensitivity and redness. These issues might impact patient adherence to treatment as well as result. Therefore, knowing on beforehand the possibility of presenting skin sensitive is useful to guiding the clinical approach, especially in regard to topical treatment.

CATEGORY	DESCRIPTION	RESULT
Predisposition to skin sensitivity	Predisposition to a more exacerbated response to medications applied topically to the skin.	**HIGH RISK**

HIGH RISK

This result indicates this patient is at high risk of skin sensitivity.
It predisposes the patient to presenting redness and sensitivity upon using topical treatment.

INDICATIONS

Low risk Medium risk High risk

3.
Predisposition to hormone-related acne

3.1 Predisposition to acne due to hormonal alteration

– HIGH RISK –

ABOUT

Hormonal profile is determined by several factors, including sex, age, nutrition, and medication intake. Nevertheless, the rate at which hormones are produced and interconverted is highly dependent on the patient's genetic profile. Therefore, a considerable part of the genetic predisposition to acne is derived from the genetic balance of hormone production.

CATEGORY	DESCRIPTION	RESULT
Predisposition to acne due to hormonal alterations	Genetic predisposition to presenting acne due to alterations in the hormonal levels, which should be treated accordingly.	**HIGH RISK**

HIGH RISK

This patient presents an increased risk of acne due to hormonal changes.
Investigation of the hormone levels should be performed to provide the best care.

INDICATIONS

Low risk Medium risk High risk

3.
Predisposition to hormone-related acne

3.1 Predisposition polycystic ovary syndrome

– HIGH RISK –

ABOUT

Polycystic ovary syndrome is a highly inherited condition affecting women, for which acne is one of the criteria involved in the diagnosis. Therefore, genetic predisposition to polycystic ovary syndrome should also be considered so it can be appropriately investigated earlier in patients predisposed to it.

Nutritional management has been found as an important tool to control the polycystic ovary syndrome. Hypocaloric diets aimed at weight loss have been shown to improve free testosterone levels, menstrual cycle alterations, acne, and overall quality-of-life.

CATEGORY	DESCRIPTION	RESULT
Predisposition to polycystic ovary syndrome	Predisposition of developing polycystic ovary syndrome, thus presenting acne	**HIGH RISK**

HIGH RISK

This patient presents an increased risk of polycystic ovary syndrome.
The investigation should be performed early to treat the condition when present.
Nutritional management, entailing a hypocaloric diet, might be useful.

INDICATIONS

Low risk Medium risk High risk

Nutritional advice

General information

Nutrition plays a vital role in the development of acne and several clinical markers related to the appearance of lesions. Lipidemia and glycemia correlate tightly to the sebaceous glands' functioning and the skin's inflammatory milieu. Maintaining a healthy and balanced diet is very relevant to mitigating the predisposition to acne and treating the condition.

Furthermore, biochemical alterations might be expected during the treatment with oral retinoids, so proper nutritional management should be indicated. In this sense, a personalized nutritional evaluation might mitigate these effects.

The nutritional plan must be, then, designed according to the patient's genetic predisposition to biochemical alterations, either pre-existing or deriving from the treatment with isotretinoin.

4.
Nutritional advice

4.1 Predisposition to retinoid-related hyperlipidemia

– HIGH RISK –

ABOUT

As retinoids bind to nuclear receptors, they alter the expression of several genes. Therefore, oral retinoid therapy might directly impact the concentration of lipoproteins circulating in the body, causing damage to health. During this therapeutic approach, some genetic markers might indicate an augmented predisposition to present hyperlipidemia. Therefore, nutritional therapy should be considered, given the risk.

CATEGORY	DESCRIPTION	RESULT
Predisposition to retinoid-related hyperlididemia	Genetic predisposition to presente higher cholesterol levels during therapy with retinoids	**HIGH RISK**

HIGH RISK

This result indicates this patient presents an increased risk of developing hyperlipidemia due to the treatment with oral retinoids.
Caution should be applied when prescribing and following this patient.

INDICATIONS

Low risk Medium risk High risk

4.
Nutritional advice

4.2 Lipid metabolism

– HIGH RISK –

ABOUT

The concentration of lipoproteins and triglycerides is highly influenced by genetics. Considering the role that these biochemical markers have in the development of acne. Proper nutritional following of patients at risk for hyperlipidemia is relevant to diminishing acne's risk and consequences.

CATEGORY	DESCRIPTION	RESULT
Lipid Metabolism	Predisposition to present hyperlipidemia regardless of retinoid therapy	**HIGH RISK**

HIGH RISK

This result indicates a higher predisposition to presenting augmented levels of cholesterol and triglycerides.

INDICATIONS

Low risk Medium risk High risk

4.
Nutritional advice

4.3 Carbohydrate metabolism

– HIGH RISK –

ABOUT

The capacity of the individual to metabolize carbohydrates is influenced by genetics. Maintaining higher glucose serum concentrations signals to several pathways, e.g., IGF-I receptor and insulin receptor, generating augmented production of sebum and inflammation in the skin as a response. Therefore, the proper nutritional recommendation is beneficial for preventing and aiding in acne treatment.

CATEGORY	DESCRIPTION	RESULT
Carbohydrate metabolism	Genetic predisposition. To presenting altered gycemia and carbohydrate metabolism	**HIGH RISK**

HIGH RISK

This patient presents an elevated risk of giving higher serum glucose levels.
Nutritional management is highly recommended.

INDICATIONS

Low risk Medium risk High risk

4. Nutritional advice

4.4 Food allergy

– HIGH RISK –

ABOUT

Food allergy often presents clinically with skin lesions due to the alteration of the immunological environment of the skin. Although food allergy does not directly cause acne, it might be connected to its manifestation due to changes in the presence of cytokines typical of the inflammatory process. Therefore, eliminating or diminishing the intake of an allergenic food might be beneficial to preventing or aiding in the treatment of acne.

CATEGORY	DESCRIPTION	RESULT
Food Allergy	Genetic predisposition to presenting food allergy, which might ellicit skin manifestations	**HIGH RISK**

HIGH RISK

This patient presents essential genetic markers of the predisposition to food allergy.
It is advised to provide nutritional counseling to determine and eliminate potential allergens.

INDICATIONS

Low risk Medium risk High risk

IV.
Complete genetic results

A list of the genotypes presented by the patient for each one of the 60 SNPs analyzed to fully understand the relevant genetic profile of that patient regarding acne.

1. Acne Predisposition

ABOUT

Here you will find the genotypes obtained for this patient to determine the predisposition to severe acne and the propensity to present consequences of those lesions.

Gene/Region	SNPiD	Transition	Risk Allele	Genotype	RISK	DESCRIPTION
IL-1B	rs16944	A>C	C	AA	**LOW**	-
Non-genic region	rs38055					
Non-genic region	rs1159268					
FST	rs629725					
TLR4	rs4986790					
TLR4	rs4986791					
MYC	rs4133274					

Gene/Region: part of the patient's DNA affected by the possible variation;
SNPID: Scientific identification for the genetic alteration;
Transition: Nucleotide alteration;
Risk allele: Nucleotide that confers a particular deleterious condition for the patient;
Genotype: Combination of nucleotides the patient presents in each copy of that gene or region
Risk: Category of risk related to that genotype
Description: a brief explanation of the phenotypic consequences related to the genotype presented by the patient

GENETIC DATA APPROVED BY
Valentina Russo, Laboratory Manager

2. Skin condition and inflammation

ABOUT

Here you will find the genotypes obtained for this patient to determine the predisposition to present skin sensitivity when using irritant agents, and the predisposition to hyperpigmentation and scars related to acne.

Gene/Region	SNPiD	Transition	Risk Allele	Genotype	RISK	DESCRIPTION
MYEF2	rs1426654	A>C	C	AA		-
TNF-α	rs1800629					
IL-10	rs1800896					
Non-genic region	rs10515088					
Non-genic region	rs763035					
CYP17A1	rs743572					
Non-genic region of the FLG	rs7927894					
HSD3B1	rs6428829					
IRF4	rs12203592					
MTA3	rs17030203					
WNT10A	rs74333950					
Non-genic region	rs873549					
FOXL2	rs1511412					
RETN	rs3745367					
RETN	rs1862513					

Gene/Region: part of the patient's DNA affected by the possible variation;
SNPID: Scientific identification for the genetic alteration;
Transition: Nucleotide alteration;
Risk allele: Nucleotide that confers a particular deleterious condition for the patient;
Genotype: Combination of nucleotides the patient presents in each copy of that gene or region
Risk: Category of risk related to that genotype
Description: a brief explanation of the phenotypic consequences related to the genotype presented by the patient

GENETIC DATA APPROVED BY
Valentina Russo, Laboratory Manager

3. Predisposition to hormone-related acne

ABOUT

Hormonal balance is of great relevance in determining: 1) inflammatory response, 2) sebum production, 3) lipid profile, and 4) glucose metabolism and insulin secretion. These might alter the onset of acne; therefore, knowing how this patient is predisposed to hormonal alterations will aid in determining the treatment.

Gene/Region	SNPiD	Transition	Risk Allele	Genotype	RISK	DESCRIPTION
CYP17A1	rs743572			AA	**LOW**	-
MYEF2	rs1426654					
CYP19A	rs700518					
FST	rs629725					
THADA	rs13429458					
THADA	rs12478601					
LHCGR	rs13405728					
FSHR	rs2268361					
FSHR	rs2349415					

Gene/Region: part of the patient's DNA affected by the possible variation;
SNPID: Scientific identification for the genetic alteration;
Transition: Nucleotide alteration;
Risk allele: Nucleotide that confers a particular deleterious condition for the patient;
Genotype: Combination of nucleotides the patient presents in each copy of that gene or region
Risk: Category of risk related to that genotype
Description: a brief explanation of the phenotypic consequences related to the genotype presented by the patient

GENETIC DATA APPROVED BY
Valentina Russo, Laboratory Manager

4. Nutritional correlation

ABOUT

Nutrition plays a vital role in determining the appearance of acne and its severity and accompanying the metabolic changes that occur during the treatment with retinoids. Furthermore, skin presentation of food allergies is a factor in modifying the skin immunologic milieu.

Gene/Region	SNPiD	Transition	Risk Allele	Genotype	RISK	DESCRIPTION
RXR	rs283696			AA	**LOW**	-
RXR	rs10918169					
RXR	rs2651860					
RXR	rs1128977					
SOAT1	rs404818					
PNPLA3	rs738409					
TM6SF2	rs58542926					
APOE	rs4420638					
ABCG8	rs6544713					
HNF1A-AS1	rs2650000					
GHRL	rs27647					
FLG-AS1	rs12123821					
IL-13	rs1295686					
C11orf30/ LRRC32	rs2212434					
SERPINB7	rs12964116					
FTO	rs8050136					
ODZ4	rs7103693					
ARAP1	rs9667947					
FABP2	rs1799883					

Gene/Region: part of the patient's DNA affected by the possible variation;
SNPID: Scientific identification for the genetic alteration;
Transition: Nucleotide alteration;
Risk allele: Nucleotide that confers a particular deleterious condition for the patient;
Genotype: Combination of nucleotides the patient presents in each copy of that gene or region
Risk: Category of risk related to that genotype
Description: a brief explanation of the phenotypic consequences related to the genotype presented by the patient

GENETIC DATA APPROVED BY
Valentina Russo, Laboratory Manager

5. Pharmacogenetics

ABOUT

Here you will find the genotypes of this patient that directly correlate to their response to some of the drugs used in the treatment of acne, both due to pharmacokinetic and pharmacodynamic factors.

The patient's genetic predisposition to respond to the main drugs involved in acne treatment significantly alters the treatment of this and other conditions. Here you will find a summary of the recommendations of this patient regarding pharmacogenetics of the essential drugs. Note that those are not necessarily drugs used in the same pharmacotherapy.

Gene/Region	SNPiD	Transition	Risk Allele	Genotype	RISK	DESCRIPTION
RXR	rs283696			AA	LOW	-
RXR	rs10918169					
RXR	rs2651860					
CYP3A5 (C__26201809_30)	rs776746					
CYP3A4*22	rs35599367					
CYP3A4*2	rs2737418					
CYP3A4*11	rs28988604					
CYP3A4*20	rs67666821					
OATP1B1	rs4149056					
ABCC2	rs717620					
HLA-DRB1	rs701829					
CYP2C9*2	rs1799853					
CYP2C9*3	rs1057910					
CYP2C9*5	rs28371686					
CYP2C9*8	rs7900194					
HLA-B*51:01	rs2442736					

Gene/Region: part of the patient's DNA affected by the possible variation;
SNPID: Scientific identification for the genetic alteration;
Transition: Nucleotide alteration;
Risk allele: Nucleotide that confers a particular deleterious condition for the patient;
Genotype: Combination of nucleotides the patient presents in each copy of that gene or region
Risk: Category of risk related to that genotype
Description: a brief explanation of the phenotypic consequences related to the genotype presented by the patient

GENETIC DATA APPROVED BY
Valentina Russo, Laboratory Manager

V.
Genetics and Acne

Basic concepts of the influence of genetics in the treatment of acne and its sequalae.

Fagron AcneTest

Fagron AcneTest is a pharmacogenomics-centered algorithm considering the genetic predisposition to skin features to guide and improve acne treatment.

Why use the Genetic approach in the treatment of acne?

Although acne is a disease commonly treated with success in the dermatological practice, the type of treatment and stage at which this approach is taken influence the outcome. Late treatment of some types of acne will make the patient prone to scar tissue formation and other long-lasting sequelae, e.g., post-inflammatory hyperpigmentation. The prescription of adequate treatment promptly is essential to achieve better results, avoiding the necessity for lengthier and costly treatments.

Despite being a frequent disease with typical onset during the teenage years, the pathogenetic aspects of acne may be strongly influenced by genetics. Approximately 81% of the biological factors related to acne are influenced by genetics. Furthermore, the genetic influence in the hormone metabolism may be part of the pathogenesis of acne in the adult woman. As an example, considering the influence of the immune response in acne, genetic variations in genes related to inflammation are essential in predicting the severity of acne and the probability of the essential sequelae.

What is evaluated in the Fagron AcneTest?

Besides a comprehensive clinical evaluation algorithm, the patient is genotyped for 60 single nucleotide polymorphisms. With that genetic profile, we generate information on 1) skin predisposition, i.e., how the patient is predisposed to acne, inflammation, scars, and hyperpigmentation; 2) pharmacogenetics, patient-specific response to medication; 3) predisposition to hormone-related acne; 4) nutritional correlation.

By genetically testing the patients, doctors are able to deeply understand underlying physiological alterations. The AcneTest allows acquiring information that would not be possible by the clinical approach. Therefore, dermatologists will be able to make better-informed decisions and provide personalized treatment.

What is pharmacogenetics?

One of the main aims of the test is to provide information on the response to drugs employed in acne treatment. For that purpose, we use the concept of pharmacogenetics. As a result, pharmacogenomics may be considered the center of personalized medicine; thus, further studying and applying pharmacogenomics leads to a better understanding of the patient and the possibility of delivering customized treatment. Furthermore, pharmacogenetic knowledge allows for better treatment adherence and improves results in refractory cases.

We may approach pharmacogenomics initially by considering two main targets: 1) variations on genes of proteins involved in the metabolism of the specific drug; 2) variations on genes of molecular targets, e.g., receptors. Considering the first target, i.e., metabolism, certain enzymes are involved in either the activation or the degradation of one or several drug molecules. Thus, genomic variants yielding more or less active enzymes will determine the pharmacokinetics of this molecule, i.e., the variation of concentration over time.

Considering the range of drugs used in acne treatment, the decision among those molecules for therapy may benefit from having precise genetic information from the patient. With that knowledge, the physician is able to choose a precise molecule and its dose. Therefore, a more effective treatment, with less side-effects is possible.

How else genetics impacts the acne treatment?

The genetic predisposition to present elevated inflammation markers is correlated to the clinical presentation of inflammatory acne and, therefore, to the sequalae following the lesions. Patients with the predisposition to inflammatory severe acne might be treated precociously so as to avoid further complications.

Some patients might also be genetically influenced to present higher glycemia or lipidemia, therefore, providing nutritional recommendation to control those biochemical parameters will aid in treating acne.

Furthermore, hormonal disbalances are key factors in the development of acne in the adult woman. Genetics allows an early understanding of how the patient metabolizes hormones and, therefore, elicits the possibility of implementing antiandrogenic treatment.

Legal disclaimer

Fagron Genomics, S.L.U carries out genetic tests upon request by healthcare professionals, in relation to biological samples from patients obtained by the healthcare professional. Our tests do not replace a medical consultation, nor do they make up a diagnostic or treatment, nor should they be interpreted this way. Only healthcare professionals can interpret the results of said tests, based on their knowledge of the clinical records of the patients and other relevant factors and, under their responsibility, give a diagnostic or prescribe treatment to the patient. We decline all responsibility derived from the use and interpretation of the results of our tests by the solicitant healthcare professional. Fagron Genomics, S.L.U expressly reserves any legal actions in case of an inappropriate, negligent or incorrect use or interpretation of the results of our tests. It is the responsibility of the healthcare professional who requests a test to guarantee to the patient the appropriate genetic advice as foreseen by Law 14/2007, of 3rd July, of biomedical research. As Fagron Genomics, S.L.U does not have access to the personal identifiable information about the patient from whom the sample comes, it is the responsibility of the requesting healthcare professional to comply with the applicable data protection Laws and regulations.

Methodology

How was this test performed?

DNA was extracted from the buccal swab sample provided and was analyzed by our clinical analysis laboratory. DNA was extracted using the KingFisher Flex® robotic extraction system (Thermo Fisher Scientific). The study of the genetic variants was carried out using a custom-designed microfluidic card to measure for the chemiluminescent detection of each of them using TaqMan® technology. TaqMan® technology for genotyping testing is proven and widely used in clinical and research settings. The sensitivity (detection limit) of this study is 99%."

We analyze 60 SNPs related to the pathogenesis, predisposition, and treatment of acne.

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