

# **Phenotype Description**

# Click on the phenotype to access its description.

Acne

Acne Controls

ADHD

Allergic rhinitis

Alzheimer's disease

Asthma

**Baldness** 

COPD

Coronary artery disease

Crohn's disease

Endometriosis

**Essential tremor** 

Glaucoma

Longevity

Macular degeneration

Major depression

Migraine

Obesity

Obsessive-compulsive disorder

Osteoarthritis

Osteoporosis

Panic disorder

**Psoriasis** 

Rheumatoid arthritis

Schizophrenia

Type 2 diabetes



#### Acne

The dermatologist or the physician had confirmed that the patient suffers or had suffered from at least one of the following forms of acne: extensive comedonal acne; extensive comedonal and papular acne; deep (cicatricial) acne with 3 or more scars; cystic acne with 3 or more cysts.

The location (head or torso) and severity of each of these forms of acne were provided. If the patient's acne was due to any of the following, then the patient was excluded from the study: medicinal acne; non-medicinal acne-like conditions (folliculitis, rosacea, perioral dermatitis, SAPHO syndrome, polycystic ovary syndrome, or hypertrichosis); occupational acne; or acne caused by use of cosmetics. The approximate age of onset and information about the medication used and the family history are also provided.

### Acne Controls

The subject were 20 years of age or older, must have never suffered from any form of acne, must have never consulted a physician or dermatologist for a problem related to acne, must have never taken anti-acne product (prescribed or over the counter) or any product to treat pimples, if a female; must have not taken birth-control pills due to acne.

Following a physical examination by a trained nurse of the face, back, sternum and shoulders, the subject must not have shown any trace of any form of acne. If a female, the subject was also asked if she suffered from acne during menstruation cycle and/or pregnancy and information about birth control medication if used before the age of 20 years old was collected. Information on family history is also provided.

#### **ADHD**

The patients were aged between 6 and 12 years old, they must have received a diagnosis of ADHD by a health professional (e.g. physician, school psychologist, psycho-educator, etc.) and a confirmed diagnosis of ADHD according to the DSM-IV criteria.

Diagnoses have been established with the French version of the Diagnostic Interview Schedule for Children - DISC-IV (computerized version) based on DSM-IVR criteria. The interview was administered to a parent by a trained interviewer and it covered the following diagnoses: anxious disorders, mood disorders, schizophrenia, disruptive behaviour disorders, substance-related disorders and other disorders usually first diagnosed in infancy, childhood or adolescence.

The cognitive functions of the child have been evaluated using the Wechsler intelligence Scale for Children – Third Edition (WISC-III). The five following sub-tests were administered to the child: information, similarities, vocabulary, digit span and block design. The arithmetic and vocabulary scales developed by Woodcock and Johnson (1990) have been used to determine the developmental age reached by the child.

The following standardized questionnaires have also been completed: Connors Behavior Inventory parent and teacher version when available, the Strengths and Weakness of ADHD symptoms Rating Scale for Parent – Swan, the Family Assessment Device – FAD, and the Temperament and Character Inventory – TCI completed by the mother and the father. These questionnaires were used to document the child symptoms and behaviours, the family environment, and parents' personality traits.



## Allergic Rhinitis

The patients must have suffered from seasonal allergic rhinitis and the diagnosis must have been confirmed by a positive cutaneous response test for ragweed, dust mites, or tree pollen. The case report form also recorded information on associated and related diseases and family history.

### Alzheimer disease

The patients must have experienced multiple cognitive deficits; memory alteration with some other symptoms such as aphasia, apraxia, agnosia or perturbation of executive functions, with an age of onset after 65 (late-onset Alzheimer's disease).

The observed cognitive deficit must be the basis of a significant alteration and decline of socioprofessional functioning and the evolution of the disease must be characterized by a progressive start and a continuous cognitive decline. If the cognitive decline can be explained by other factors such as Parkinson's disease, Huntington's disease, cerebrovascular disease, brain tumours, schizophrenia, major depression, etc. or if the cognitive deficit is induced by a substance, then the patient is excluded. The results of cognitive tests (MMSE and MoCA) were recorded, when available, as well as the family history.

#### **Asthma**

The patients must have suffered from asthma as diagnosed by a physician, either a pneumologist or an allergist with an age of onset before 35 years old. Bronchodilatation measurements and/or allergy test results are recorded. A qualitative assessment of severity of the symptoms and exacerbations, presence of associated and related diseases, and family history are also included.

#### **Baldness**

The patients were males with a stage VI or VII baldness on the Norwood-Hamilton scale (confirmed by photographs prior to age 40). The onset of the condition must have occurred prior to age 25 and the condition must not be from an infectious, psychological, toxic, cicatricial, or hormonal origin. The age at which the patient had hair patterns corresponding to stage I and IV on the Norwood-Hamilton scale for the last time is noted. Medication, response to treatment, if any, and family history were also recorded.

#### COPD

The patients must have had a current diagnosis of COPD (including chronic bronchitis and/or emphysema) at recruitment, exhibited obstructive syndrome (FEV1/FVC <70%), suffered appearance of symptoms after the age of 30, been a smoker (minimum of 10 pack years) and not been suffering from any of the following disorders: asthma, allergic rhinitis, lung cancer, tuberculosis, cystic fibrosis of the pancreas, bronchiectasis and pulmonary resection. The case report form also included information on associated symptoms, smoking habits, further diagnostic information (e.g. FEV/FVC, arterial blood gas measures), treatment and family history for both COPD and asthma.



### **Coronary Artery Disease**

The patients must have had a diagnosis of coronary disease before the age of 55 for a man or before the age of 60 for a woman, exhibited a BMI of 35 or lower, not been suffering from diabetes and exhibited an HDL cholesterol level lower than the 10<sup>th</sup> percentile. The lipid measurements values are provided. The form also provides information on lipid-lowering medication, prior events of myocardial infarction, coronary bypass or angioplasty and alcohol drinking habits.

Other patients with coronary artery disease, myocardial infarction, coronary bypass or angioplasty have also been selected from the participants with broad consent of the biobank.

### Crohn's disease

The diagnosis of Crohn's disease must have been obtained during a colonoscopy, radiological examination with barium, abdominal surgical operation, or from tissue diagnosis (biopsy or surgical specimen). Patients also suffering from ulcerative or infectious colitis and from other intestinal diseases were excluded. The case report form also recorded clinical information on the associated symptoms, bowel areas affected and the clinical expression, hospitalization and surgery, presence of associated diseases, treatment and family history.

### **Endometriosis**

The diagnosis of endometriosis must have been confirmed by the presence of typical black lesions found during a surgical procedure, laparoscopy or from a pathology report. The disease stage I, II, III or IV as defined by the American Society for Reproductive Medicine (ASRM) was recorded. The following information was also recorded: presence of chronic pelvic pain, dysmenorrhea, dyspareunia and infertility, reason for surgery, history of fertility and family history.

### **Essential Tremor**

The patient must have suffered from a bilateral action tremor (postural or kinetic) of the hands and forearms, or had isolated head tremor without apparent signs of dystonia. Moreover, the patient must be free of many other neurologic or associated diseases including Parkinson's disease, dystonia and peripheral neuropathy. The form also included the age of onset, family history, medication information and the results of a full-body examination for presence of tremor.

### Glaucoma

The patients must have had a diagnosis of open-angle glaucoma with an onset after the age of 40. Comorbidity with macular degeneration is noted. Various measurements as indication of the severity of the disease are collected, including angle measurements and intraocular tension measurements following the Goldmann test. Medication and family history were also collected.



## Longevity

The subjects must have been aged 95 years or older. The case report form recorded general health information, life style habits, medication and nutrient supplements. The results of a cognitive test (MMSE) were also part of the available dataset.

### **Macular Degeneration**

The patients must have had a diagnosis of macular degeneration of the wet form as confirmed following an eye examination by a competent physician and a photograph of the retina. The age of onset and some data on lifestyles were collected. Symptoms associated with macular degeneration, visual acuity and clinical manifestations were surveyed. The family history was also collected.

### **Major Depression**

The patients must have met the diagnostic criteria for major depressive disorder, recurrent and severe with or without psychotic features according to DSM-IV. The case report form provided information on the psychotic features, if applicable, medication and family history.

### Migraine

The patient must have suffered from migraine with aura following the criteria of the IHS (International Headache Society, section I, II and III). The form included information on the nature and duration of the aura symptoms, and the factors initiating the symptoms (food, environment, stress and hormones).

# Obesity

The patients must have exhibited a body mass index (BMI)  $\geq$  30. Information on the presence of associated diseases, qualitative assessment of blood dosages, dietary information and family history were also recorded. Other patients with obesity have also been selected from the participants with broad consent of the biobank.

# Obsessive-compulsive Disorder

The patients must have had a confirmed primary diagnosis of obsessive-compulsive disorder according to DSM-IV criteria. The patients were diagnosed following a SCID-I interview and the specific types of obsessions and compulsions are documented with the administration of the Yale-Brown Obsessive-Compulsive Symptom Checklist.

The patients were also asked to complete the following related questionnaires; SCID-I Personality Disorder Questionnaire, Severity of Alcohol Dependence Data (SADD), Severity of Dependence Scale (SDS), Beck Depression Inventory (BDI-II), Maudsley Obsessive-Compulsive Inventory (MOCI), Padova Inventory and a Questionnaire for Distinguishing Obsessive-Compulsive Disorder with Tourette's Syndrome.



### Osteoarthritis

The patients must have suffered from hand osteoarthritis and be aged 60 years or less or to have been clinically (or by X-ray) diagnosed for hand osteoarthritis before 60 years of age. A standard hand osteoarthritis workup, i.e. postero-anterior radiographs of the hands were performed if not already done in the last year prior to recruiting. The medical files included the radiographs.

# Osteoporosis

All patients were female and must have had non-traumatic vertebral fractures, a low bone density score (T <-1.5), been in menopause for at least one year and never taken steroids (Prednisone  $\geq$  7.5 mg/day> 3 months). Information on the fractures history, pregnancy history, data from radiology and biochemical tests, medication and family history were also provided.

For this disease, a case report form has also been completed for the controls. To qualify as a control, a woman must have never had a vertebral fracture or a non-traumatic fracture after 49 years of age, been in menopause for at least one year; never taken steroids (Prednisone ≥ 7.5 mg/day> 3 months) and not shown a low bone density (Score T ≥-1.5). Complementary information on some diseases, medication and family history are also available.

### Panic Disorder

The patients must have a confirmed primary diagnosis of panic disorder with or without agoraphobia, or a diagnosis of agoraphobia without a history of panic disorder, according to DSM-IV criteria. Diagnoses of panic disorder and/or agoraphobia, other anxiety disorders, somatoform disorders and eating disorders have been established with the Structured Clinical Interview for DSM-IV Axis-I disorders administered by trained interviewers. The patients were also asked to complete the following related questionnaires; Anxiety Sensitivity Index, Mobility Inventory for Agoraphobia, Body Sensation Questionnaire and Agoraphobic Cognition's Questionnaire as well as life situation questionnaires.

### **Psoriasis**

The patients must have exhibited the criteria for diagnosis of plaque forming psoriasis (psoriasis vulgaris) with an age of onset between 18 and 40. The various body areas affected were recorded, as well as medication and treatment history and the presence of comorbid disorders.

### **Rheumatoid Arthritis**

The phenotype criteria follow those of the American College of Rheumatology (ACR) and the diagnosis must have been performed at least 2 years prior to recruitment. In addition, the age of onset must have been after the age of 18 with a documented test for positive serum rheumatoid factor or with presence of erosions on hands or feet documented through X-rays. The form provided further information on the presence of extraarticular features, test for anti-CCP, presence of other autoimmune disorders, medication and family history.



## Schizophrenia

The phenotype was a clinician-based diagnosis following the DSM-IV criteria. The following information was captured on the case report form: disease subtype (paranoid, disorganized, catatonic, undifferentiated, or residual), qualitative assessments of the severity of the positive, negative and cognitive symptoms, hospitalization and placement history, healthcare services required, dysfunctions exhibited by the patient, presence of comorbid disorders as defined by DSM-IV, abuse of psychotic substances, medication and family history.

### Type 2 Diabetes

The diagnosis of type 2 diabetes was based on the criteria established by the Canadian Diabetes Association, with an onset after 30 years old and a body mass index (BMI)>25. Glycaemia measurements must have been either one of the following: random glycaemia ≥11.1 mmol/L; fasting glycaemia ≥7.0 mmol/L or glycaemia during the induction of hyperglycaemia of ≥11.1 mmol/L. The following information was also recorded: lipid measurements before treatment, if known, presence of associate associated diseases, treatment and medication history and family history.