



## Overview of 23andMe Phenotype Data Collection

The majority of 23andMe's phenotypic data is collected through online questionnaires and "Quick Questions" (single, unrelated questions). There are now over 55 questionnaires and over 500 Quick Questions available on the 23andMe website. The questionnaires are described below.

### Covariates

- Where Are You From?
  - 22 questions
  - Where you, your parents, and your grandparents were born.
  - What other ethnic identities did they have (e.g., Ashkenazi, Basque, Cherokee).
  
- Health Habits
  - 45 questions
  - Covers basic common covariates
  - Height
  - Weight
  - Birth weight
  - Number of children
  - Number of young children
  - Amount and type of exercise
  - Caffeine, tobacco, and alcohol use past and present
  - Other recreational drug use
  
- What Do You Do?
  - 9 questions
  - Basic socio-economic covariates
    - Occupation
    - Employment status
    - Marital status
    - Earnings
    - Amount of time spent working
    - Education
  
- Ten Things About Your Environment
  - 22 questions
  - Covers common environmental risk factors for several diseases
    - Use of herbicide
    - Use of pesticide
    - Amount of fish eaten
    - Source of tap water and type of filter used
    - Mold in house

- Smoking
  - Exposure to secondhand smoke
  - Residence near farms
  - Residence near factories
  - Zip code (urban/rural/suburban)
- Your Physical Activity
  - 41 questions
  - Adapted from the International Physical Activity Questionnaire (IPAQ), Craig et al. 2003. "International physical activity questionnaire: 12-country reliability and validity." *Med Sci Sports Exerc* 35(8):1381-95.
  - Measures amount of vigorous, moderate, and other activity over the past week.
- Under Pressure
  - 10 questions
  - Perceived stress scale from Cohen et al. (1983) . "A global measure of perceived stress." *J Health Soc Behav* 24(4):385-96.
- Caffeine Usage
  - 57 questions
  - Covers duration and frequency of use of coffee, black tea, green tea, and caffeinated sodas
- Pesticides at Work
  - 182 questions
  - Covers occupational exposure to pesticides
    - Type of pesticide
    - Name of pesticide
    - Age at exposure
    - Duration of exposure
    - Type of exposure
    - Use of protective equipment
    - Adverse reactions to pesticide exposure
- Smoking
  - 24 questions
  - Use of tobacco products
    - Duration and amount
- Head Injury
  - 52 questions
  - Incidence of head injury or concussion
  - Number of injuries
  - Severity
    - Hospitalization
    - Loss of consciousness

- Amnesia, trouble thinking
- Seizures
- Skull fracture

## Medical

### Allergies and Asthma

- Allergies and Asthma
  - 250 questions
  - Allergies from:
    - Food
    - Plants
    - Animals
    - Dust mites
    - Mold
    - Latex medications
    - Vaccines
    - Other
  - In each category, follow-up questions about
    - What food/plant/etc. causes the allergy
    - Whether it was diagnosed by a doctor
    - Age of allergic reaction
    - Type of reaction
  - Severe allergy attacks
  - Asthma attacks
  - Triggers for asthma
  - Number of older/younger siblings
  - Pets as a child
  - Maternal allergies while pregnant
  - Family history
- Asthma
  - 20 questions
  - Age of diagnosis, current status
  - Allergy symptoms
  - Frequency of shortness of breath
  - Frequency of waking due to asthma symptoms
  - Frequency of use of rescue inhaler
  - Triggers (animals, smoke, dust, pollen, food, etc.)
  - FEV1 result from spirometry
  - Past, current usage and efficacy of Singulair
  - Other medication usage

### Baldness

- Hair Loss in Men and Women
  - 20 questions

- Hair loss as compared to a set of pictures, age at beginning of hair loss
- Conditions possibly causing hair loss (giving birth, medications, anemia, PCOS, etc.)
- Medications used for hair loss (minoxidil, finasteride, etc.)
- Efficacy of these medications

## **General Medical**

- Your Medical History
  - 384 questions
  - A comprehensive medical history.
  - Asks if you have had 191 different diseases, conditions, and surgeries, and if so, at what age were you diagnosed.
  - Full list of conditions covered can be found at the end of the Questionnaires section.
- Cancer Personal and Family History
  - 120 questions
  - Assesses personal history of cancer and family history of cancer in 1<sup>st</sup> degree relatives and grandparents

## **H1N1**

- H1N1 Flu (Swine Flu)
  - 29 questions
  - Covers symptoms, severity, and response to antivirals for people who had H1N1 flu

## **Healthy Aging**

- Longevity
  - 26 questions
  - Age at death (or current age) for yourself, parents, and grandparents
  - Cause of death (accidental versus non) for parents and grandparents
- Sports Injuries
  - 138 questions
  - Activities engaged in on a regular basis (from aerobics to martial arts to yoga)
    - Age ranges when you participated in these activities
    - Time on a job involving manual labor
    - Time using a computer daily
    - ACL tears (when, from which activity)
    - Meniscus tears
    - Tendinitis (achilles, elbow, patellar, other; from what activity)
    - Rotator cuff tears
    - Frequent or chronic knee or shoulder pain

## **Infertility**

- Male Infertility
  - 53 questions
  - Number of offspring, impregnation
  - Time to conception

- Infertility treatment
- Semen analyses
- Infertility diagnosis
- Risk factors
  - Wet heat exposure
  - Cycling
  - Diseases affecting fertility
  - Fertility-related surgeries
- Female Infertility
  - 45 questions
  - Number of pregnancies
  - Time to first conception
  - Age at first pregnancy or trying to conceive
  - Infertility treatment
  - Infertility diagnosis
  - Miscarriages
  - Risk factors
    - Diseases affecting fertility
    - Fertility-related surgeries

## **Medication Response and Side Effects**

- Commonly Used Medications
  - 41 questions
  - Efficacy of OTC medications
    - Second-generation antihistamines
    - Acetaminophen
    - NSAIDS (aspirin, ibuprofen, Aleve)
    - Codeine
    - PPIs (omeprazole, etc.)
  - Side effects of OTC and common prescription medications
    - Second-generation antihistamines and dry mouth, fast heartbeat
    - First-generation antihistamines and dizziness / drowsiness
    - Pseudoephedrine and jitter
    - NSAIDS and stomach problems
    - Codeine and euphoria, constipation, vomiting, itching
    - Niacin (with or without a statin) and flushing
- Finasteride
  - 95 questions
  - Use of medications for hair loss: duration and amount
  - Degree of hair loss before and after treatment
  - Self-rated efficacy
- NSAIDs
  - 548 questions

- Use of NSAIDs: duration and amount
  - Experience of side effects
  - Efficacy
- Warfarin
  - 224 questions
  - Use of warfarin: dosage and duration
  - Dosage changes
  - Reason prescribed
  - Side effects
- Antidepressants
  - 259 questions
  - History of antidepressant use
  - Duration and amount
  - Indication
  - Side effects
  - Effectiveness for depression
- Proton Pump Inhibitors
  - 136 questions
  - History of proton pump inhibitor use
  - Duration of use
  - Indication
  - Side effects
  - Efficacy
- Serious Side Effects
  - 8 questions
  - Collects information about severe drug side effects
- Parkinson's disease, migraines, sarcoma, MPN background, and baldness surveys also include questions about drug response

## **Migraines**

- Migraines
  - 63 questions
  - Detailed survey about causes and symptoms of migraines
    - Frequency and duration of headaches
    - Symptoms during a migraine
    - Preceding warning signs
    - Visual changes
    - Family history
    - Medications used (NSAIDS, dihydroergotamine, ergotamine, metoclopramide, butalbital combinations, opiates, triptans)
    - Efficacy of these medications
    - Prophylactic medications (beta blockers, calcium channel blockers, antihypertensives, tricyclic antidepressants, SSRIs, anti-seizure drugs, cyproheptadine)
    - Possible triggers

## **Myeloproliferative Neoplasms**

- Background Survey
  - 41 questions
  - Diagnosis
  - Age diagnosed
  - Diagnosing physician
  - Confirmation by bone marrow biopsy
  - Presence of germline mutations
  - Risk factors
    - Family history of MPNs
    - Exposure to petroleum chemicals
- MPN Symptoms
  - 62 questions
  - Includes the Myelofibrosis Symptom Assessment Form, developed by Ruben A. Mesa
  - Assesses symptoms in the last week
    - Pain
    - Early satiety
    - Fevers
    - Fatigue
    - Night sweats
    - Itching
  - Blood counts
    - Hemoglobin
    - White blood cells
    - Peripheral blasts
  - Weight change
  - Stroke or blood clots
  - Treatment for clots
  - Bleeding

- Diagnosis and treatment of gout

## **Parkinson's Disease**

- Parkinson's Disease: Background
  - 104 questions
  - Created in collaboration with the Parkinson's Institute and other PD experts
  - Comprehensive survey about Parkinson's disease (PD)
  - Covers:
    - Diagnosis
      - When and by what type of doctor?
      - Diagnosis of other comorbid conditions that may be confused with PD (PSP, MSA, essential tremor, etc.)
      - Initial onset of PD (initial symptoms, sudden/gradual onset, unilateral/bilateral, progression of symptoms, improvement with medication)
    - For people without a diagnosis of PD:
      - Assessment of movement characteristics (tremor, handwriting, balance, etc.) that are associated with PD
    - Medications
      - Medications that may cause PD like symptoms (e.g., certain psychiatric medications)
      - Medications currently and previously taking for PD (Sinemet, Stalevo, and 8 others)
      - Efficacy of medications on a typical day
    - For people with a diagnosis of PD
      - Age at which specific symptoms began to show up
      - Universal Parkinson's Disease Rating Scale (UPDRS), rating how severe symptoms have been over the past week
      - Whether they had a caregiver fill out the survey.
    - Environmental risk factors
      - Family history of PD
      - Smoking
      - Pesticide use on the job
      - Pesticide use at home
      - Head injuries
      - NSAID use
- Parkinson's Disease: Tracking
  - 38 questions
  - Longitudinal collection of:
    - Symptoms
      - UPDRS
    - Medications
      - Which drugs
      - Efficacy



- Supplements
- Surgeries/treatments
  - Deep brain stimulation
  - Other surgeries
- Therapies
  - Occupational / counseling / speech / etc.
- Exercise
  - Amount of vigorous / moderate activity in the last week
- Meant to be taken every few months

### **Post-operative Nausea and Vomiting (PONV)**

- After-Effects of Surgery
  - 38 questions
  - Covers surgeries under general anesthesia and the extent of nausea or vomiting afterwards, as well as any family history of PONV

### **Pregnancy**

- Weekly Pregnancy Survey
  - 40 week longitudinal survey, to be taken each week while pregnant
- Past Pregnancy and Birth Survey
  - 266 questions
  - Gathers a detailed retrospective history for all pregnancies
  - Covers conception, prenatal care, multiple births, miscarriages, weight gain, symptoms, diseases during pregnancy, changes in chronic conditions, early delivery, delivery method, complications, birth weight, postnatal complications

### **Sarcoma**

- Sarcoma Background Survey
  - 287 questions covering
  - Diagnosis and subtype
  - Diagnosing and treating physician types
  - Symptoms
  - Disease course and treatment
  - Treatment efficacy
  - Side effects from treatments
  - Risk factors for sarcoma
    - Military service
    - Prior cancer, metastasis
    - Radiation therapy
    - Exposure to Agent Orange
    - Family history of cancer, sarcoma

### **Urinary Incontinence**

- Urinary Incontinence Questionnaire
  - 27 questions

- Stress and urge urinary incontinence
- Medications for incontinence
- Surgery for incontinence
- Effectiveness of treatment
- Pelvic organ prolapse
- Risk factors
  - Vaginal deliveries
  - Chemotherapy

## Traits

### Laterality

- Ocular Dominance
  - 1 question
  - A simple test to measure which eye is dominant
- Handedness
  - 12 questions
  - Asks about which hand is dominant in a variety of motions (writing, throwing, shoveling, etc.) in order to score handedness on an 8 point scale.
  - Survey from Annett M (1970). "A classification of hand preference by association analysis." Br J Psychol 61(3):303-21.
- Footedness
  - 12 questions
  - Which foot is dominant for a variety of motions.
  - Elias et al. (1998). "Footedness is a better predictor than is handedness of emotional lateralization." Neuropsychologia 36(1):37-43.

### Morphology

- Physical Features
  - 37 questions about:
    - Upper, lower back hair
    - Cheek, chin dimples
    - Tongue flip, eyebrow lifting
    - Toe, finger lengths
    - Shoe size(s), arch height
    - Bra band and cup size
    - 11 questions from the Beighton hypermobility scale
    - Face shape, nose profile, brow line
- Skin aging
  - 60 questions, limited to women with age > 30 and no cosmetic surgery
  - Forehead, under-eye, next-to-eye, brow wrinkles
  - Facial sagging
  - Lip height

- Age spots
  - Sun burns, tanning ability
  - Smoking history
  - Hormone exposure
    - Use of birth control pills
    - Pregnancy status
    - Menopausal status
    - Hormone replacement therapy
  - Sun exposure
    - Country, state of residence in childhood, adulthood
    - Outdoor employment
    - Tanning bed use
    - Sunbathing frequency
    - Sunscreen use, rating
  - Use of medications affecting sun sensitivity:
    - Accutane, retinoic acid
    - Celebrex, Vioxx
    - Statins
  - Use of facial skin care products with:
    - Alpha-hydroxy acids
    - Moisturizer, with or without sunscreen
    - Retinol, retinoids
    - Salicylic acid
  - Skin cancer
  - Diagnoses of diabetes, hyperglycemia
- Rosacea
    - 13 questions, limited to men and women 30 and older
    - Symptoms of rosacea affecting the face:
      - Persistent redness
      - Broken blood vessels
      - Persistent papules (small red bumps)
      - Cobbled appearance of nose
      - Prolonged flushing
      - Burning or stinging
      - Swelling
      - Gritty feeling / dry eyes
    - Diagnosis
    - Diagnosis of other autoimmune conditions
    - Treatment for rosacea

### **Miscellaneous Traits**

- Ten Things About You
  - 10 quick questions about:
  - Photic sneeze reflex
  - Night/morning person

- Hand clasp
- Wear glasses
- Cavities
- Handedness
- Freckles
- Preference for sweet food
- Asparagus anosmia
- Hair curl
- Ten More Things About You
  - 10 more quick questions about:
  - Attached earlobes
  - Braces
  - Wisdom teeth
  - Motion sickness (in cars, planes, boats)
  - Leg jiggle
  - Hair whorl
  - Astigmatism
  - Eyelid crease

## Personality

- Optimism
  - 10 questions
  - Measures optimism on a 25 point scale
  - From Scheier et al. (1994). "Distinguishing optimism from neuroticism (and trait anxiety, self-mastery, and self-esteem): a reevaluation of the Life Orientation Test." J Pers Soc Psychol 67(6):1063-78.
- Five Dimensions of Personality
  - 44 questions
  - Measures personality on five factors: neuroticism, extraversion, openness, agreeableness, and conscientiousness
  - From John, O. P. and Srivastava, S. (1999) . "The big-five trait taxonomy: History, measurement, and theoretical perspectives." New York: Guilford.
- Reading the Mind in the Eyes-Revised Version
  - 38 questions
  - Assesses the ability to read others' emotional states component of empathy
  - Developed by Simon Baron-Cohen at the Autism Research Centre

## Pigmentation

- Pigmentation
  - 11 questions
  - Eye color, hair color, and freckling at age five and at adulthood.
  - Pictures and text descriptions used to classify hair color into 9 categories, eye color into 7 categories, and freckling on a 16 point scale.

- Skin Color
  - 11 questions
  - Skin color and tanning response using pictures and descriptions.

## **Conditions covered in the general health survey**

### **Addiction**

- Alcoholism
- Drug abuse
- Nicotine abuse

### **Bone and joint**

- Chronic back pain
- Degenerative joint disease
- Fracture
- Joint hyperextensibility (double-jointedness)
- Osteoarthritis
- Osteoporosis
- Scoliosis

### **Cancer**

- Anal cancer
- Basal cell carcinoma
- Bladder cancer
- Bone cancer
- Brain cancer
- Breast cancer
- Cervical cancer
- Colorectal cancer
- Endometrial cancer
- Esophageal cancer
- Gall bladder cancer
- Kidney cancer
- Leukemia
- Liver cancer
- Lung cancer
- Melanoma
- Mouth cancer
- Non-cancerous breast mass
- Non-Hodgkin's lymphoma
- Ovarian cancer
- Pancreatic cancer

- Prostate cancer
- Salivary gland cancer
- Squamous cell carcinoma
- Stomach cancer
- Testicular cancer
- Throat cancer
- Thyroid cancer
- Uterine cancer
- Vaginal cancer
- Vulval cancer

### **Gastrointestinal**

- Blood in your stool
- Celiac disease
- Chronic constipation/Hirschsprung's disease
- Colon polyps
- Crohn's disease
- Gastroesophageal reflux disease (acid reflux disease)
- Heartburn
- Hemorrhoids
- Hernia
- Hiatal hernia (when your stomach pushes into your esophagus)
- Intestinal polyps
- Ulcer
- Ulcerative colitis

### **Heart**

- Angina (chest pain treated with nitroglycerin)
- Aortic aneurysm
- Aortic stenosis
- Congenital heart defect
- Coronary artery disease/heart disease
- Heart attack
- Heart failure
- Myocarditis (inflammation of the muscular part of the heart)

### **Immune**

- Asthma
- Drug allergy
- Environmental (but not seasonal) allergy
- Food allergy
- Lupus
- Rheumatoid arthritis

- Seasonal allergies

### **Kidney**

- Bladder infection
- Kidney cysts
- Kidney stones
- Reduced kidney function (requiring dialysis)

### **Liver**

- Cirrhosis
- Elevated result on liver function test
- Hemochromatosis
- Hepatitis
- Liver cysts

### **Lung**

- Chronic bronchitis
- Emphysema
- Pneumonia
- Positive response (red bump) to tuberculosis (TB) skin test, patch test, PPD
- Tuberculosis

### **Mental health**

- Anxiety disorder
- Attention deficit disorder (ADD) or Attention deficit hyperactivity disorder (ADHD)
- Autism-spectrum disorder
- Bipolar disorder (manic depression)
- Depression
- Dissociative identity disorder (multiple personality disorder)
- Eating disorder (anorexia, bulimia)
- Mental retardation or other learning difficulties
- Obsessive-compulsive disorder
- Post-traumatic stress disorder
- Schizophrenia

### **Metabolic and circulatory**

- Anemia
- Bleeding tendency
- Blood clots
- Blood disorder
- Gestational diabetes
- High blood pressure
- High blood sugar (hyperglycemia)
- High cholesterol (over 200mg/dl) or Hypercholesterolemia
- Orthostatic hypotension
- Stroke
- Type 1 diabetes

- Type 2 diabetes

### **Neurological**

- Alzheimer's disease
- Dyslexia
- Loss of sensation
- Memory loss
- Migraine headaches with aura
- Migraine headaches without aura
- Multiple sclerosis
- Paralysis
- Parkinson's disease
- Recurrent vertigo (dizziness)
- Seizure disorder (epilepsy)
- Traumatic brain injury

### **Other**

- AIDS or HIV+
- Chicken pox
- Chronic fatigue syndrome
- Cold sores
- Fibromyalgia
- Measles
- Mumps
- Obesity
- Polio
- Rheumatic fever
- Rubella
- Scarlet fever
- Shingles

### **Reproductive**

- Endometriosis
- Infertility
- Ovarian cyst
- Prostate enlargement

### **Sensory**

- Astigmatism
- Blindness
- Cataract
- Farsightedness (far objects are clear, near objects are blurry)
- Glaucoma
- Hearing loss



- Macular degeneration
- Nearsightedness (near objects are clear, far objects are blurry)
- Retinal detachment/retinal tears
- Ringing in the ears (tinnitus)

### **Sleep**

- Sleep apnea
- Sleep disturbance

### **Skin**

- Eczema
- Psoriasis
- Rosacea
- Scleroderma
- Several large moles (1/4 inch, flat or raised) or abnormal biopsy of mole
- Severe acne
- Skin tags
- Three or more severe sunburns that produced blisters
- Urticaria (chronic hives)

### **Surgical procedures**

- Angioplasty
- Appendectomy
- Arthroscopic surgery
- Biopsy (not breast)
- Bone fracture repair
- Brain surgery
- Breast augmentation
- Breast biopsy
- Breast reduction
- Bunionectomy
- Cardiac ablation (to treat arrhythmia)
- Cataract removal
- Cervical spine surgery
- Cesarean section (C-section)
- Coronary bypass surgery
- Corrective eye surgery (not LASIK)
- Cosmetic/plastic surgery
- Ear tube insertion/myringotomy
- Gall bladder removal
- Gastrectomy (partial or full removal of the stomach)
- Gastric bypass surgery (stomach stapling)
- Heart valve surgery

- Hemorrhoid surgery
- Hernia repair
- Hysterectomy (removal of the uterus)
- Joint replacement
- Joint surgery
- Laparoscopy
- LASIK
- Liposuction
- Lumbar spine surgery
- Lumpectomy
- Mastectomy
- Mohs surgery (for basal cell carcinoma)
- Open heart surgery
- Tonsillectomy (surgery to remove the tonsils)
- Transplant surgery

### **Thyroid**

- Hyperthyroidism
- Hypothyroidism
- Thyroid nodules