

**This is a sample of the GenomeConnect Initial Health Survey. If you are interested in participating in GenomeConnect, you will complete the health survey in your registry account at [genomeconnect.org](https://genomeconnect.org).**

# GenomeConnect - Health Survey

## How do you identify your gender/sex?

- Female
- Male
- Intersex
- MTF/Transwoman/Transgender Female
- FTM/Transman/Transgender Male
- Unknown
- Other

## Have you had genetic testing?

Examples: Chromosome microarray (CMA single gene sequencing), single gene test, gene panel test, whole exome sequencing (WES), whole genome sequencing (WGS)

- Yes
- No
- Unsure

## Genetic Testing

### What type(s) of genetic testing have you had? Please select all that apply.

- Chromosomes (Also called chromosome analysis, karyotype, karyotype analysis, or G-band karyotype)
- Direct-to-consumer (DTC) genetic test (Genetic testing that is ordered by and returned directly to the person tested rather than through a doctor, also called DTC; please specify name of the company and/or test below)
- Single gene or gene panel test (Testing of ONE or MULTIPLE genes with a single test; please specify the name of the test or condition being tested for below)
- Microarray (Also called chromosomal microarray, CMA, cytogenomic array, array, or array CGH)
- Exome sequencing (Testing that targets a person's complete set of genes; also called whole exome sequencing or WES)
- Genome sequencing (Testing of a person's complete DNA sequence, including genes and regions between them; also called whole genome sequencing or WGS)
- Unsure

### What was the reason for your genetic testing?

- I have had symptoms of a genetic condition.
- The doctor/I wanted to confirm a diagnosis that was suspected based on my symptoms.
- I have a family history of a genetic disorder and was showing symptoms of that disorder – wanted to confirm diagnosis.
- I have a family history of a genetic disorder and was NOT showing symptoms of the disorder – wanted to assess my risk. I am healthy and wanted to be proactive about my health.
- Unsure

**Do you have a copy of your genetic test result(s)?**

\*\*Instructions for uploading a copy of your genetic test report(s) will be provided after you complete this survey. If you do not yet have a copy of your genetic test(s), you can contact the GenomeConnect coordinator to get help locating and submitting a copy of the report.

- Yes (I have had testing and have a copy.)
- No (I have had testing but do NOT have a copy.)
- Unsure

**Health & Development**

**Have you been told that there were issues during your biological mother's PREGNANCY with you and/or your DELIVERY/BIRTH?**

Examples: Mother had to be put on bed rest, there were concerns on ultrasound, you were born early (prematurely), or you had to stay in the neonatal intensive care unit (NICU).

- Yes
- No
- Unsure

**Have you had issues with your GROWTH?**

Examples: You are much shorter or taller than expected for your family, you have had significant and/or lifelong problems gaining weight, or you have significant and/or lifelong problems with obesity. You may have seen an endocrinologist or other specialist for these issues.

- Yes
- No
- Unsure

**Have you had issues with your HORMONES and/or ENDOCRINE SYSTEM (hormone-producing glands)?**

Examples: Diabetes, issues with estrogen or testosterone levels, or function of the thyroid, parathyroid, or adrenal glands. You may have seen an endocrinologist for these issues.

- Yes
- No
- Unsure

**Have you ever been diagnosed with cancer?**

Examples: Breast cancer, prostate cancer, osteosarcoma, lymphoma, etc. You may have seen an oncologist for these issues.

- Yes
- No
- Unsure

**Have you had issues with your HEAD/FACE/NECK?**

Please note that we are asking you about SIGNIFICANT issues with these areas, problems that you have seen a doctor for or had surgery for, or problems that you don't notice often in other people. The doctor may have used the term "dysmorphic" in describing some features of your face.

Examples: Cleft lip/palate, large or small head size, fused skull bones, sparse hair, etc. We will ask

specifically about eyes and ears later.

- Yes
- No
- Unsure

**Have you had issues with your EYES and/or VISION?**

Examples: Vision loss, cataracts, a “lazy eye,” nearsighted- or farsightedness. You may have seen an optometrist or ophthalmologist (eye doctors) for these issues.

- Yes
- No
- Unsure

**Have you had issues with your EARS and/or HEARING?**

Examples: Hearing loss, ringing in the ears, or misshapen ears. You may have seen an audiologist or Ears/Nose/Throat (ENT) doctor for these issues.

- Yes
- No
- Unsure

**Have you had issues with BRAIN and/or NERVOUS SYSTEM (neurological issues)?**

Examples: Developmental delays, intellectual disability, learning problems, seizures, significant problems with muscle tone, cerebral palsy, or problems with brain structure. You may have seen a neurologist for these issues.

- Yes
- No
- Unsure

**Have you had BEHAVIOR and/or PSYCHIATRIC issues?**

Examples: Autism spectrum disorders, schizophrenia, anxiety, aggression, repetitive behaviors, and depression. You may have seen a developmental pediatrician, psychologist, psychiatrist, or therapist for these issues.

- Yes
- No
- Unsure

**Have you had issues with your SKIN?**

Examples: Unusual skin coloration, lumps or bumps that needed to be looked at by a doctor, or sweating issues. You may have seen a dermatologist for these issues.

- Yes
- No
- Unsure

**Have you had issues with your BONES, CARTILAGE, and/or CONNECTIVE TISSUE?**

Examples: Issues with easily broken bones, extreme flexibility, or the look/use of your arms, legs, hands, and/or feet. You may have seen an orthopedist or other special doctor for these issues.

- Yes

- No
- Unsure

**Have you had issues with your MUSCLES?**

Examples: Significant issues with the way your muscles are formed or the way they work, including muscle weakness, burning, or stiffness. You may have seen a neurologist, orthopedist, or other special doctor for these issues.

- Yes
- No
- Unsure

**Have you had issues with your HEART and/or BLOOD VESSELS?**

Examples: Cardiomyopathy, heart defects needing surgery, widened/bulging blood vessel, or problems with heart rhythm. You may have seen a cardiologist for these issues.

- Yes
- No
- Unsure

**Have you had issues with your LUNGS and/or BREATHING?**

Examples: A collapsed lung, fluid on the lungs, or apnea. You may have seen a pulmonologist for these issues.

- Yes
- No
- Unsure

**Have you had issues with your DIGESTIVE SYSTEM?**

Examples: Includes stomach, intestines, or other parts of gastrointestinal (GI) tract. Examples: Issues with the structure or use of your stomach or intestines, extreme issues with constipation, reflux, and/or vomiting. You may have seen a GI (gastrointestinal) doctor for these issues.

- Yes
- No
- Unsure

**Have you had issues with your KIDNEYS, BLADDER, and/or GENITALS (male/female reproductive parts)?**

Examples: Issues with urination, kidneys, or reproductive system, including concerns about how these body parts look or work. Some specific examples are hypospadias, kidney failure, infertility, and incontinence. You may have seen a nephrologist, urologist, gynecologist, or other special doctor for these issues.

- Yes
- No
- Unsure

**Have you had issues with your IMMUNE SYSTEM?**

Examples: Frequent infections or immunodeficiency. You may have seen an immunologist for these issues.

- Yes

- No
- Unsure

**Have you had issues with your BLOOD and/or BLEEDING?**

Examples: Excessive bleeding or problems noted with your blood cells on lab reports. You may have seen a hematologist for these issues.

- Yes
- No
- Unsure

**Have you had issues with your ORAL HEALTH?**

- Yes
- No
- Unsure

The following sections are assigned based on answers to initial questions.

**Mother's Pregnancy**

**What specific issues were there during your biological mother's PREGNANCY with you and/or your DELIVERY/BIRTH?**

Answers	HPO Term	Time of symptoms	Time of diagnosis
<b>Amniotic fluid issue</b> - Too much fluid (polyhydramnios) or too little fluid (oligohydramnios) in the amniotic sac	Abnormality of the amniotic fluid; HP:0001560	<ul style="list-style-type: none"> <li>• First trimester</li> <li>• Second trimester</li> <li>• Third trimester</li> <li>• At delivery</li> <li>• Unsure</li> </ul>	<ul style="list-style-type: none"> <li>• First trimester</li> <li>• Second trimester</li> <li>• Third trimester</li> <li>• At delivery</li> <li>• Unsure</li> </ul>
<b>Decreased fetal movement</b> - Baby moved less than expected during pregnancy	Decreased fetal movement; HP:0001558	<b>Repeated for each field</b>	<b>Repeated for each field</b>
<b>Delivery complication</b> - Can include breech birth, forceps or vacuum delivery, or umbilical cord wrapped around the baby's neck	Abnormal delivery, HP:0001787		
<b>Maternal health problem</b> - An issue with the mother's health during pregnancy; can include issues such as high blood pressure, diabetes, or CMV infection, etc.	Prenatal maternal abnormality; HP:0002686		
<b>Placental issue</b> - Placenta was not formed, attached, or working properly	Abnormality of the placenta; HP:0100767		
<b>Pregnancy exposure</b> - Mother had an exposure that could cause harm to the baby, like alcohol, cigarettes, street drugs,	Maternal teratogenic		

chemicals, or radiation, etc.	exposure; HP:0011438		
<b>Prematurity</b> - Baby was born early, before 37 weeks gestation	Premature birth; HP:0001622		
<b>Umbilical cord issue</b> - Cord was not formed properly or had an issue like a knot	Abnormality of the umbilical cord; HP:0010881		

**Are there any other specific issues during your biological mother's PREGNANCY with you and/or your DELIVERY/BIRTH that were not listed above? Please include at what age did the issues begin, and when were they diagnosed by a healthcare provider.**

**Sometimes babies who are born at full term or close to term can have complications or health problems after birth, just like many premature babies. Were you born at "term" (between 37-40 weeks) and experience health complications after birth?**

If you were born preterm (a "preemie," born before 37 weeks), skip over this question. If your birth/delivery occurred at 37 weeks or later, and you required no special nursery care, please mark "No" here.

- Yes
- No
- Unsure

### Growth

**What specific GROWTH issues have you had? At what age did the GROWTH issues begin, and when were they diagnosed by a healthcare provider?**

Answers	HPO Term	Age symptoms first appeared	Age at diagnosis
<b>General overgrowth</b> - Significant and/or lifelong large size or overgrowth, often due to a syndrome	Generalized overgrowth; HP:0001548	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul> <p><b>Repeated for each field</b></p>	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul> <p><b>Repeated for each field</b></p>
<b>Obesity</b> - Significant and/or lifelong issues with weight not specifically related to an overgrowth syndrome	Obesity; HP:0001513		
<b>Uneven growth</b> - One side of the body is larger or longer than the other side (hemihypertrophy)	Hemihypertrophy; HP:0001528		
<b>Short stature</b> - Height is much shorter than is typical for your family and/or the average height for someone of your same age and sex	Short stature; HP:0004322		
<b>Undergrowth</b> - Significant problems growing as expected during childhood (failure to thrive)	Failure to thrive; HP:0001508		

<b>Growth hormone deficiency</b> - Insufficient production of growth hormone from pituitary gland	Growth hormone deficiency; HP:0000824		
<b>Growth hormone excess</b> - Too much production of growth hormone from pituitary gland; can lead to acromegaly	Growth hormone excess; HP:0000845		
<b>Tall stature</b> - Height much taller than is typical for your family and/or the average height from someone of your same age and sex	Tall stature; HP:0000098		

**Are there any other specific GROWTH issues you've had that were not listed above? Please include at what age did the GROWTH issues begin, and when were they diagnosed by a healthcare provider.**

### Hormone/Endocrine

**What specific HORMONES/ENDOCRINE issues have you had? At what age did the HORMONE/ENDOCRINE issues begin, and when were they diagnosed by a healthcare provider?**

Answers	HPO Term	Age symptoms first appeared	Age at diagnosis
<b>Type 2 diabetes</b> - The most common form of diabetes, sometimes called adult, late-onset, or non-insulin dependent; may or may not require insulin	Non-insulin dependent diabetes; HP:0005978	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul>	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul>
<b>Type 1 diabetes</b> - Insulin-dependent diabetes, sometimes also called juvenile or autoimmune diabetes; typically requires insulin	Insulin dependent diabetes; HP:0100651	<ul style="list-style-type: none"> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul>	<b>Repeated for each field</b>
<b>Delayed puberty</b> - Passing the age when puberty normally occurs without any signs of physical or hormonal puberty	Delayed puberty; HP:0000823	<ul style="list-style-type: none"> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul>	
<b>Diabetes insipidus</b> - Less-common form of diabetes showing extreme thirst and frequent urination	Diabetes insipidus; HP:0000873	<b>Repeated for each field</b>	
<b>Growth hormone deficiency</b>	Growth hormone deficiency; HP:0000824		
<b>Early puberty</b> - Puberty before the age of 8 years in girls or 9 years in boys; also called precocious puberty	Precocious puberty; HP:0000826		
<b>Hypogonadism</b> - Little or no sex hormones produced by the testes in men or the ovaries in women	Hypogonadism; HP:0000135		

<b>Adrenal hyperplasia</b> - Enlargement of the adrenal gland leading to decreased production of the hormone cortisol	Adrenal hyperplasia; HP:0008221		
<b>Hyperthyroidism</b> - High levels of thyroid hormone, including Graves' disease	Thyroid goiter HP:0000853		
<b>Hypothyroidism</b> - Low levels of thyroid hormone	Hyperthyroidism; HP:0000836		
<b>Goiter</b> - Enlargement of the thyroid gland which may result in neck swelling	Hypothyroidism; HP:0000836		
<b>Parathyroid issue</b> - Too much or too little parathyroid hormone produced	Parathyroid dysfunction; HP:0011767		

Are there any other specific HORMONES/ENDOCRINE issues you've had that were not listed above? Please include at what age did the HORMONES/ENDOCRINE issues begin, and when were they diagnosed by a healthcare provider.

### Cancer

Please choose the answer that best describes the kind of cancer you had and how old you were when diagnosed. If you do not see the type you had, please check 'Other'.

### Answers

Breast Cancer	Breast carcinoma; HP:0003002	<b>Age at diagnosis</b> <ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul> <b>Repeated for each field</b>
Ovarian Cancer	Ovarian neoplasm; HP:0100615	
Uterine cancer	Uterine Neoplasm; HP:0010784	
Cervix	Cervix cancer; HP:0030079	
Prostate	Prostate neoplasm; HP:0100787	
Lung	Neoplasm of the lungs; HP:0100526	
Colon and rectum (including polyps)	Colon cancer; HP:0003003	

Pancreas	Neoplasm of the pancreas; HP:0002894	
Bladder	Bladder neoplasm; HP:0009725	
Kidney	Renal neoplasm; HP:0009726	
Thyroid	Neoplasm of the thyroid gland; HP:0100031	
Melanoma of skin	Melanoma; HP:0002861	
Non-melanoma of skin	Neoplasm of the skin; HP:0008069	
Brain or spine	Neoplasm of the central nervous system; HP:0100006	
Blood (including leukemia and lymphoma)	Abnormality of the blood and blood-forming tissues; HP:0001871	
Unsure		
Other		

**Have you had any other type of cancer that was not listed above? Please include at what age was the cancer diagnosed by a healthcare provider.**

### Head/Face/Neck

**What specific issues have you had with your HEAD/FACE/NECK? At what age did the HEAD/FACE/NECK issues begin, and when were they diagnosed by a healthcare provider?**

Please select all that apply. We have listed out the different parts of the head, face, and neck that may be different, in terms of its appearance or how it works. When we ask if you have had problems with your teeth, we are NOT asking about the occasional cavity or the need for braces.

Answers	HPO	Age symptoms first appeared	Age at diagnosis
<b>Chin</b> - Can include differences in shape or structure of chin, such as an underdeveloped or recessed appearance, etc.	Abnormality of the chin, HP:0000306	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> </ul>	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> </ul>
<b>Cleft lip and/or cleft palate</b> - A "split" or division in the lip and/or roof of the mouth present at birth	Oral cleft; HP:0000202	<ul style="list-style-type: none"> <li>• 0 - 3 months old</li> <li>• 4 - 7 months</li> </ul>	<ul style="list-style-type: none"> <li>• 0 - 3 months old</li> <li>• 4 - 7 months</li> </ul>

<b>General face shape</b> - Facial features may have been described as atypical or have a shape/look that differs from the rest of the family	Abnormal facial shape; HP:0001999)	<b>Repeated for each field</b>	<b>Repeated for each field</b>
<b>Hair</b> - Can include having too little or "sparse" hair or a hair texture different from what is expected, etc.	Abnormality of the hair; HP:0001595		
<b>Inside of the mouth</b> - Can include issues with inside of the mouth, teeth, gums, or tongue	Abnormality of the oral cavity; HP:0000163		
<b>Lips or outside of the mouth</b> - Can include issues with lip or mouth size, shape, or overall development	Abnormality of the mouth; HP:0000153		
<b>Neck</b> - Can include a short neck, broad neck, webbed neck, branchial clefts, branchial pits, or branchial cysts, etc.	Abnormality of the neck; HP:0000464		
<b>Nose</b> - Broad category that can include a significant problem with the way the nose was formed or the way that it works	Abnormality of the nose; HP:0000366		
<b>Skull</b> - The bones in the head and/or face developed differently; can include a large head size (macrocephaly), small head size (microcephaly), or problems with the way the bones formed or fused (craniosynostosis), etc.	Abnormality of the skull; HP:0000929		
<b>Throat/swallowing difficulties</b> - Also called oral pharyngeal dysphagia	Oral pharyngeal dysphagia; HP:0200136		

**Are there any other specific HEAD/FACE/NECK issues you've had that were not listed above? Please include at what age did the HEAD/FACE/NECK issues begin, and when were they diagnosed by a healthcare provider.**

**Eyes/Vision**

**What specific EYE/VISION issues have you had? At what age did the EYE/VISION issues begin, and when were they diagnosed by a healthcare provider?**

<b>Answers</b>	<b>HPO</b>	<b>Age symptoms first appeared</b>	<b>Age at diagnosis</b>
<b>Eye movement issues</b> - Eyes have unusual movement such as "lazy eye," (strabismus) or eye tremors (nystagmus), etc.	Abnormality of eye movement; HP:0000496	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul>	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> </ul>
<b>Difference in eye size</b> - Eye size differs from what is usual such as a small eye size (microphthalmia) or lack of eye development (anophthalmia), etc.	Abnormality of globe size; HP:0100887		
<b>Unusual Iris</b> - The colored part of the eye, called the iris, has freckles/spots or eye color differs between eyes.	Abnormality of the iris; HP:0000525		

<b>Coloboma</b> – A notched pupil	Coloboma; HP:0000589	<b>Repeated for each field</b>	<ul style="list-style-type: none"> <li>• 69 years old</li> <li>• 70 and older</li> </ul> <b>Repeated for each field</b>
<b>Lens issue</b> - A cataract or dislocated lens (ectopia lentis), etc.	Abnormality of the lens; HP:0000517		
<b>Major vision issues</b> - Issues may include blindness, color blindness, night vision problems, floaters, or light sensitivity, etc.; do NOT include minor nearsightedness or farsightedness.	Abnormality of vision; HP:0000504		
<b>Farsightedness</b> - Also called hyperopia or hypermetropia; things up close look fuzzy	Myopia; HP:0000545		
<b>Nearsightedness</b> - Also called myopia; things in the distance look fuzzy	Hypermetropia; HP:0000540		
<b>Optic nerve issues</b> - Optic nerve is located behind the eyeball and brings information from the eye to the brain, allowing you to see; can include issues with the way this nerve was formed or the way it works	Abnormality of the optic nerve; HP:0000587		
<b>Retina issues</b> - The retina is the layer of tissue at the back of the inner eye, and the macula is included in this category; can include issues such as retinal detachment, macular	Abnormality of the retina; HP:0000479		
<b>Ptosis</b> - Droopiness of the upper eyelid	Ptosis; HP:0000508		

**Are there any other specific EYE/VISION issues you've had that were not listed above? Please include at what age did the EYE/VISION issues begin, and when were they diagnosed by a healthcare provider.**

### Ears/Hearing

**What specific EAR/HEARING issues have you had? At what age did the EAR/HEARING issues begin, and when were they diagnosed by a healthcare provider?**

Answers	HPO	Age symptoms first appeared	Age at diagnosis
<b>Ear shape difference</b> - Part or all of the ear may be over- or under-developed; can include issues such as an ear structure that is missing or underdeveloped, a cupped ear shape, or an overly-large appearance of the ear, etc.	Abnormality of the ear; HP:0000598	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> </ul>	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul>
<b>Conductive hearing loss</b> - Hearing loss due to an issue with the ear canal, ear drum, or the bones in the middle ear	Conductive hearing impairment; HP:0000405	<ul style="list-style-type: none"> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul>	

<b>Sensorineural hearing loss (SNHL)</b> - Hearing loss due to an issue with the nerves in the inner ear or connecting ear to the brain	Sensorineural hearing impairment; HP:0000407	<b>Repeated for each field</b>	<b>Repeated for each field</b>
<b>Mixed hearing loss</b> - A combination of both conductive and sensorineural hearing loss	Mixed hearing impairment; HP:0000410		
<b>Hearing loss, unsure which type</b>	Hearing impairment; HP:0000365		
<b>Ring in the ears</b> - Also called tinnitus	Tinnitus; HP:0000360		
<b>Hyperacusis</b> - Oversensitivity to or difficulty tolerating certain sounds	Hyperacusis; HP:0010780		
<b>Dizziness</b> - Also called vertigo	Vertigo; HP:0002321		

Are there any other specific EARS/HEARING issues you've had that were not listed above? Please include at what age did the EARS/HEARING issues begin, and when were they diagnosed by a healthcare provider.

#### Brain/Nervous System

What specific BRAIN/NERVOUS SYSTEM issues have you had? At what age did the BRAIN/NERVOUS SYSTEM issues begin, and when were they diagnosed by a healthcare provider?

Answers	HPO	Age symptoms first appeared	Age at diagnosis
<b>Brain and/or spinal cord issue</b> - Includes problems with the formation or appearance of the brain and/or spinal cord, often found by MRI	Abnormality of the nervous system; HP:0000707	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul>	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul>
<b>Cerebral palsy (CP)</b> - A single damaging event to the brain that can cause movement problems, seizures, learning problems, and/or communication problems	Cerebral Palsy; HP:0100021		
<b>Cognitive impairment</b> - Significant difficulties learning, understanding, or processing information, or making decisions	Cognitive impairment; HP:0100543		
<b>Coordination issues</b> - Significant clumsiness or other difficulties with movement due to body parts not working together as they normally would	Abnormality of coordination; HP:0011443		
<b>EEG abnormality</b> - A problem has been identified on an electroencephalogram, or "EEG," a test that measures the electrical activity of the brain	EEG abnormality; HP:0002353		
<b>Encephalopathy</b> - Damage to the brain, sometimes ongoing, that results in altered	Encephalopathy; HP:0001298		
		<b>Repeated for each field</b>	<b>Repeated for each field</b>

brain function			
<b>Hypertonia</b> - Muscle tightness that limits movement, spasticity, increased muscle tone	Hypertonia; HP:0001276		
<b>Hypotonia</b> - Unusually low muscle tone, muscle tone may have been described as floppy	Hypotonia; HP:0001290		
<b>Memory impairment</b> - Increased forgetfulness or significant difficulty remembering things	Memory impairment HP:0002354		
<b>Unusual movements</b> - Changes or problems in the ability to move body parts voluntarily; do NOT include Parkinson's Disease here	Abnormality of movement; HP:0100022		
<b>Parkinsonism</b> - Abnormal movements, characterized by shaking, stiffness, slowness, and difficulty walking	Parkinsonism; HP:0001300		
<b>Seizures</b> - Also known as convulsions	Seizures; HP:0001250		

Are there any other specific BRAIN/NERVOUS SYSTEM issues you've had that were not listed above? Please include at what age BRAIN/NERVOUS SYSTEM issues began, and when were they diagnosed by a healthcare provider.

#### Behavior

What specific BEHAVIORAL/PSYCHIATRIC issues have you had? At what age did the BEHAVIORAL/PSYCHIATRIC issues begin, and when were they diagnosed by a healthcare provider?

Answers	HPO	Age symptoms first appeared	Age at diagnosis
<b>Autism spectrum disorder (ASD)</b> - Includes autism and pervasive developmental disorder, not otherwise specified (PDD-NOS)	Autism Spectrum Disorder; HP:0000729	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul>	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul>
<b>Bipolar disorder</b> - Also known as manic depression; characterized by severe mood swings that affect the ability to perform day-to-day activities	Bipolar affective disorder; HP:0007302	<b>Repeated for each field</b>	<b>Repeated for each field</b>
<b>Schizophrenia</b> - A severe disorder in which people have difficulty determining what is real and unreal, and have unusual social behaviors	Schizophrenia; HP:0100753		
<b>Repetitive or self-injurious behavior</b> - Can include issues such as body-rocking, head-banging, hand-wringing, or skin picking, etc.	Stereotypic behavior; HP:0000733		
<b>Aggressive/violent behavior</b>	Aggressive/violent behavior; HP:0006919		
<b>Anxiety</b> - Feelings of dread or fear that interfere with daily living	Anxiety; HP:0000739		

<b>Depression</b>	Depression; HP:0000716		
<b>Delusions</b> - Believing that something is true despite evidence showing it is not	Delusions; HP:0000746		
<b>Hallucinations</b> - Seeing or hearing things that are not real while you are awake	Hallucinations; HP:0000738		
<b>Obsessive compulsive disorder (OCD)</b> - The need to perform recurrent rituals that interfere with your daily life	Obsessive compulsive disorder; HP:0000722		
<b>Psychosis</b>	Psychosis; HP:0000709		
<b>Short attention span</b> - Includes attention deficit disorder (ADD) and attention deficit hyperactivity disorder (ADHD)	Short attention span; HP:0000736		

Are there any other specific **BEHAVIORAL/PSYCHIATRIC** issues you've had that were not listed above? Please include at what age did the **BEHAVIORAL/PSYCHIATRIC** issues begin, and when were they diagnosed by a healthcare provider.

#### Skin

What specific **SKIN** issues have you had? At what age did the **SKIN** issues begin, and when were they diagnosed by a healthcare provider?

Answers	HPO Term	Age symptoms first appeared	Age at diagnosis
<b>Atrophic scars</b> - Scars that retract or indent	Atrophic scarring; HP:0001075	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> </ul>	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> </ul>
<b>"Café-au-lait" spots</b> - Light brown birth marks the color of coffee with milk	Multiple café-au-lait spots; HP:0007565	<ul style="list-style-type: none"> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> </ul>	<ul style="list-style-type: none"> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> </ul>
<b>Hyperpigmentation</b> - Patches of skin that are darker than the regular skin color	Hyperpigmentation; HP:0000953	<ul style="list-style-type: none"> <li>• 1 year old</li> <li>• ...</li> </ul>	<ul style="list-style-type: none"> <li>• 1 year old</li> <li>• ...</li> </ul>
<b>Hypopigmentation</b> - Patches of skin that are LIGHTER than the regular skin color	Hypopigmentation; HP:0001010	<ul style="list-style-type: none"> <li>• 69 years old</li> <li>• 70 and older</li> </ul>	<ul style="list-style-type: none"> <li>• 69 years old</li> <li>• 70 and older</li> </ul>
<b>Decreased sweating</b> - Also called hypohidrosis; you are not able to sweat or your ability to sweat is significantly reduced	Hypohidrosis; HP:0000966	<b>Repeated for each field</b>	<b>Repeated for each field</b>
<b>Increased sweating</b> - Also called "hyperhidrosis"; your sweating is significantly increased, above what is necessary to maintain a normal body temperature	Hyperhidrosis; HP:0000975		
<b>Skin fragility</b> - Skin is fragile to touch and can include issues like	Fragile skin; HP:0001030		

ectodermal dysplasia			
<b>Skin sensitivity</b> - Skin is fragile to sunlight	Skin photosensitivity; HP:0000992		
<b>Thick skin</b> - Sometimes called keratoderma or hyperkeratosis; skin in some regions of the body is significantly thickened, often occurring on the palms of the hands or soles of the feet	Thick skin; HP:0001072		
<b>Hyperelastic skin</b> - Skin that is extremely stretchy and returns to its original position after stretching	Hyperelastic skin; HP:0000974		
<b>Hypoelastic skin</b> - Inelastic or "saggy" skin, often hanging in loose folds (cutis laxa)	Cutis laxa; HP:0000973		

**Are there any other specific SKIN issues you've had that were not listed above? Please include at what age did the SKIN issues begin, and when were they diagnosed by a healthcare provider.**

#### **Bone/Cartilage/Connective Tissue**

**What specific BONE/CARTILAGE/CONNECTIVE TISSUE issues have you had? At what age did the BONE/CARTILAGE/CONNECTIVE TISSUE issues begin, and when were they diagnosed by a healthcare provider?**

<b>Answers</b>	<b>HPO Term</b>	<b>Age symptoms first appeared</b>	<b>Age at diagnosis</b>
<b>Arm and/or leg bone differences since birth</b> - Can include differences in any limb, such as a shortened arm or leg length, or the bones of the arm or leg that are formed differently, etc.	Abnormality of limb bone morphology; HP: 0002813	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul>	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul>
<b>Curvature of the spine</b> - Spine curves side-to-side (scoliosis) or front-to-back (kyphosis)	Abnormal curvature of the spine; HP:0010674	<b>Repeated for each field</b>	<b>Repeated for each field</b>
<b>Flexible joints</b> - Commonly called being double-jointed, hypermobile, or hyper extensible	Joint hyperextensibility; HP:0001382		
<b>Frequent broken bones</b> - Bones tend to break with minimal trauma, includes "brittle bone disease"	Increased susceptibility to fractures; HP: 0002659		
<b>Finger and/or toe differences</b> - Includes bony differences such as extra, missing, or fused fingers and/or toes, etc.	Abnormality of the digits; HP:0011297)		

<b>Hand differences</b> - Includes bony differences such as "split" hand or extra or missing bones in the hand.	HP:0001155		
<b>Foot differences</b> - Includes bony differences such as "split" foot or extra or missing bones in the foot.	HP:0001760		
<b>Hip dysplasia</b> - Issues with the formation of the hip socket, typically present at birth	Hip dysplasia; HP:0001385		
<b>Pectus carinatum</b> - Breastbone sticks out from the chest, sometimes called "pigeon chest"	Pectus carinatum; HP:0000768		
<b>Pectus excavatum</b> - Breastbone appears sunken or caved in to the chest, sometimes called "funnel chest"	Pectus excavatum; HP:0000767		
<b>Skeletal dysplasia</b> - Issues or differences with the growth of the bones and/or cartilage, sometimes called dwarfism	Skeletal dysplasia; HP:0002652		

**Are there any other specific BONE/CARTILAGE/CONNECTIVE TISSUE issues you've had that were not listed above? Please include at what age did the BONE/CARTILAGE/CONNECTIVE TISSUE issues begin, and when were they diagnosed by a healthcare provider.**

### Muscles

**What specific MUSCLE issues have you had? At what age did the MUSCLE issues begin, and when were they diagnosed by a healthcare provider?**

Answers	HPO Term	Age symptoms first appeared	Age at diagnosis
<b>Facial muscle issue</b> Issues with the muscles of the face; can include facial palsy, difficulty with chewing, or problems moving your eyes, etc.	Abnormality of facial musculature; HP:0000301	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul>	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul>
<b>Issue with muscle function</b> Issues with the muscles of the face; can include facial palsy, difficulty with chewing, or problems moving your eyes, etc.	Abnormality of muscle physiology; HP:0011804	<p><b>Repeated for each field</b></p>	<p><b>Repeated for each field</b></p>
<b>Issue with internal muscle structure</b> Issues with the way the muscles are formed; often diagnosed by a muscle biopsy	Abnormality of muscle morphology; HP:0011805		
<b>Issue with nerves connected to muscles</b> Issues with the way nerves communicate with muscles; often diagnosed with nerve conduction studies or electromyography (EMG)	Abnormality of the somatic nervous system: HP:0410009		

<b>Muscle issues in the arms and/or legs</b> Weakness or underdevelopment	Abnormality of the musculature of the limbs; HP:0009127)		
<b>Muscle issues in the chest, back, and/or shoulders</b> Weakness or underdevelopment	Abnormality of the musculature of the thorax; HP:0009131		
<b>Neck muscle issue</b> Torticollis or neck weakness	Abnormality of the musculature: HP:0003011		

**Are there any other specific MUSCLE issues you've had that were not listed above? Please include at what age did the MUSCLE issues begin, and when were they diagnosed by a healthcare provider.**

### Heart/Blood Vessels

**What specific HEART/BLOOD VESSEL issues have you had? At what age did the HEART/BLOOD VESSEL issues begin, and when were they diagnosed by a healthcare provider?**

Answers	HPO Term	Age symptoms first appeared	Age at diagnosis
<b>Aneurysm</b> - An enlarged or bulging blood vessel	Aneurysm; HP:0002617	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul> <p><b>Repeated for each field</b></p>	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul> <p><b>Repeated for each field</b></p>
<b>Arrhythmia</b> - A heart rhythm that is too fast, slow, or irregular	Cardiac arrhythmia; HP:0011675		
<b>Abnormal EKG (ECG)</b> - Issues with the electrical signals of the heart, seen on electrocardiogram	Abnormal EKG; HP:0003115		
<b>Arterial dissection</b> - A tear or rip in a large blood vessel	Arterial Dissection; HP:0005294		
<b>Cardiomyopathy</b> - Issues with the heart muscle; can include enlargement, thickening, or scarring	Cardiomyopathy; HP:0001638		
<b>Fainting</b> - Passing out (syncope)	Abnormality of the cardiovascular system; HP:0001626		
<b>Heart defect</b> - Issues with the way the heart and/or vessels were formed, often diagnosed at birth	Malformation of the heart or great vessels; HP:0002564		
<b>High blood pressure</b> - Also called hypertension; if this is known to be related to another condition you have, like pregnancy or pheochromocytoma, please provide more information below	Hypertension; HP:0000822		
<b>High cholesterol</b> - High levels of cholesterol in the blood, also called hypercholesterolemia	Hypercholesterolemia; HP:0003124		

<b>Stroke</b> - Loss of blood supply to the brain	Stroke; HP:0001297		
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**Are there any other specific HEART/BLOOD VESSEL issues you've had that were not listed above? Please include at what age did the HEART/BLOOD VESSEL issues begin, and when were they diagnosed by a healthcare provider.**

### Lungs/Breathing

**What specific LUNG/BREATHING issues have you had? At what age did the LUNG/BREATHING issues begin, and when were they diagnosed by a healthcare provider?**

Answers	HPO Term	Age symptoms first appeared	Age at diagnosis
<b>Asthma</b>	Asthma; HP:0002099	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul>	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul>
<b>Chronic lung issues</b> - Issues with the shape, size, or health of the lung; these issues are on-going and you should NOT include occasional illnesses like respiratory infections or pneumonia	Abnormality of the respiratory system; HP:0002086	<b>Repeated for each field</b>	<b>Repeated for each field</b>
<b>Decreased lung function</b> - Issues with lungs working at "full capacity"; also called decreased pulmonary function	Decreased pulmonary function; HP:0005952		
<b>Diaphragm issues</b> - Issues with the muscle that separates your chest from your abdomen such as congenital diaphragmatic hernia or weakness of the diaphragm	Abnormality of the diaphragm; HP:0000775)		
<b>Respiratory insufficiency</b> - Issues with taking in enough oxygen and/or getting rid of enough carbon dioxide	Respiratory insufficiency; HP:0002093		
<b>Restrictive lung disease</b> - Issues with fully expanding the lungs with air; can be caused by issues outside of the lungs such as neuromuscular disorders, scoliosis, or sarcoidosis	Restrictive lung disease; HP:0002091)		
<b>Unusual breathing patterns</b> - Issues include apnea, hyperventilation, or rapid breathing (tachypnea)	Abnormal pattern of respiration; HP:0002793		
<b>Upper respiratory tract issues</b> - Issues with the voice box (larynx), windpipe (trachea), or air tubes leading to lungs (bronchi)	Abnormality of the upper respiratory tract; HP:0002087		

**Are there any other specific LUNG/BREATHING issues you've had that were not listed above? Please include at what age did the LUNG/BREATHING issues begin, and when were**

they diagnosed by a healthcare provider.

## Digestive System

What specific DIGESTIVE SYSTEM issues have you had, or what part is affected? At what age did the DIGESTIVE SYSTEM issues begin, and when were they diagnosed by a healthcare provider?

Answers	HPO Term	Age symptoms first appeared	Age at diagnosis
<b>Feeding difficulties</b> - Issues with eating; can include problems eating food by mouth or keeping food from being vomited	Feeding difficulties; HP:0011968	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul>	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul>
<b>Esophagus</b> - Issues with the tube that takes food from your mouth to your stomach; can include reflux, inflammation, or narrowing of the esophagus, etc.	Abnormality of the esophagus; HP:0002031	<p><b>Repeated for each field</b></p>	<p><b>Repeated for each field</b></p>
<b>Stomach</b> - Issues with the stomach; can include a hernia, pyloric stenosis, or problems emptying food from the stomach, etc.	Abnormality of the stomach; HP:0002577		
<b>Pancreas</b> - Issues with pancreas such as pancreatitis or cysts	Abnormality of the pancreas; HP:0001732		
<b>Liver</b> - Issues with liver; can include enlargement (hepatomegaly), scarring (cirrhosis), hepatitis, or cysts, etc.	Abnormality of the liver; HP:0001392		
<b>Dysmotility</b> - Issues moving food through the digestive system; can include ileus or intestinal pseudo-obstruction, etc.; do NOT include conditions like Crohn's disease or colitis in this category	Gastrointestinal dysmotility; HP:0002579		
<b>Intestines</b> - Issues with the small intestine, large intestine, bowel, and colon; can include malabsorption, intestinal malrotation, polyps, diverticulitis, Crohn's disease, or colitis, etc.	Abnormality of the intestine; HP:0002242		
<b>Soiling/defecation</b> - Issues with producing a normal bowel movement (feces/poop); can include bowel incontinence, colitis, malrotation of the colon, etc.	Abnormality of the large intestine; 0002250		
<b>Anus</b> - Issues with the opening between the buttocks through which solid waste (feces/poop) leaves the body such as narrowing of the anal opening or absence of the anus at birth; sometimes called anorectal malformation	Abnormality of the anus; HP:0004378		

Are there any other specific DIGESTIVE SYSTEM issues you've had that were not listed above? Please include at what age did the DIGESTIVE SYSTEM issues begin, and when were they diagnosed by a healthcare provider.

### Kidney/Bladder/Genitals

What specific KIDNEY/BLADDER/GENITAL issues have you had? At what age did the KIDNEY/BLADDER/GENITAL issues begin, and when were they diagnosed by a healthcare provider?

Answers	HPO Term	Age symptoms first appeared	Age at diagnosis
<b>Bladder</b> - Issues with how the bladder is shaped or works; can include problems with bladder stones, bladder size, overactive bladder, or incontinence, etc.	Abnormality of the bladder; HP:0000014	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul>	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul>
<b>Abnormal internal or external sex organ</b> - Also called disorder of sex development (DSD); can include having both male and female reproductive organs, having external genitalia that is not clearly male or female, or having sex organs that do not go through normal changes at puberty	Abnormal sex determination; HP:0012244	<ul style="list-style-type: none"> <li>• 69 years old</li> <li>• 70 and older</li> </ul>	<ul style="list-style-type: none"> <li>• 69 years old</li> <li>• 70 and older</li> </ul>
<b>Kidney function issue</b> - Issue with how the kidneys work such as kidney failure or problems with proper filtering/absorption	adrenal hyperplasia; HP:0008221	<p><b>Repeated for each field</b></p>	<p><b>Repeated for each field</b></p>
<b>Kidney structure issue</b> - Issue with the kidney shape/structure; can include abnormal size, missing kidney, or abnormal kidney location in the body	Kidney malformation; HP:0012210		
<b>Female sex organ disorder</b> - Issues with the formation or appearance of the female genitalia, including labia, vagina, uterus, and/or ovaries	Abnormality of FEMALE genitalia; HP:0010460		
<b>Genitalia function</b> - Issues with the way the male or female genitalia works; can include infertility, impotence, or ovarian failure, etc.	Genital functional abnormality; HP:0000080		
<b>Male sex organ disorder</b> - Issues with the formation or appearance of the male genitalia, including penis, scrotum, testicles, and/or prostate gland	Abnormality of MALE genitalia; HP:0010461		
<b>Urethra issue</b> - Issues with the tube that empties the bladder; can include problems with way the urethra formed, blockage, or abnormal urethra connection (hypospadias), etc.	Abnormality of the urethra; HP:0000795		

<b>Ureter issue</b> - Issues with tube connecting the kidney to the bladder; can include ureter size being too large or small, abnormal ureter formation or blockage, or missing ureter, etc.	Abnormality of the ureter; HP:0000069		
<b>Urine ("pee") issues</b> - Issues with urine; can include blood or protein in urine (hematuria or proteinuria), unusual urine color, or unusual urine odor, etc.	Abnormality of the urine; HP:0003110		

SAMPLE

## GenomeConnect Survey

Are there any other specific KIDNEY/BLADDER/GENITAL issues you've had that were not listed above? Please include at what age did the KIDNEY/BLADDER/GENITAL issues begin, and when were they diagnosed by a healthcare provider.

### Immune System

What specific IMMUNE SYSTEM issues have you had? At what age did the IMMUNE SYSTEM issues begin, and when were they diagnosed by a healthcare provider?

\*Please note that we are not asking about common allergies (such as allergies to foods, animals, or the environment) in this question.\*

Answers	HPO Term	Age symptoms first appeared	Age at diagnosis
<b>Autoimmune disorder</b> - Disorder when the immune system attacks the body's own healthy tissues; can include Hashimoto thyroiditis, lupus, or autoimmune thrombocytopenia, etc.	Autoimmunity; HP:0002960	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> </ul>	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul>
<b>Immunodeficiency</b> - Lack of an effective immune response to fight an infection or illness; can include T-cell immunodeficiency, other cellular immunodeficiencies, or combined immunodeficiency, etc.	Immunodeficiency; HP:0002721	<ul style="list-style-type: none"> <li>• 69 years old</li> <li>• 70 and older</li> </ul> <p><b>Repeated for each field</b></p>	<p><b>Repeated for each field</b></p>
<b>Increased inflammatory response</b> - An overreaction of the body to injury or infection that results in too much redness or swelling	Increased inflammatory response; HP:0012647		
<b>Recurrent infections</b> - Infections that "come back" more often than normal despite treatment	Recurrent Infections; HP:0002719		
<b>Rheumatoid arthritis</b> - An autoimmune-caused form of arthritis; includes juvenile and adult forms	Rheumatoid arthritis; HP:0001370		

Are there any other specific IMMUNE SYSTEM issues you've had that were not listed above? Please include at what age did the IMMUNE SYSTEM issues begin, and when were they diagnosed by a healthcare provider.

### Blood/Bleeding

## GenomeConnect Survey

**What specific BLOOD/BLEEDING issues have you had? At what age did the BLOOD/BLEEDING issues begin, and when were they diagnosed by a healthcare provider?**

These options are listed showing what type of cell has issues or what type of blood issue occurs. You can select more than one for the same issue.

Answers	HPO Term	Age symptoms first appeared	Age at diagnosis
<b>Blood clot</b> - Select this choice if a <u>blood clot has occurred</u> in one or more of your veins or arteries (thrombosis)	Abnormal thrombosis; HP:0001977	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul>	<ul style="list-style-type: none"> <li>• Unsure</li> <li>• Before birth</li> <li>• 0 - 3 months old</li> <li>• 4 - 7 months old</li> <li>• 8 - 11 months old</li> <li>• 1 year old</li> <li>• ...</li> <li>• 69 years old</li> <li>• 70 and older</li> </ul>
<b>Blood clotting</b> - Select this choice if you have a <u>coagulation disorder</u> , meaning an issue with your blood clotting too fast or too slow	Abnormality of coagulation; HP:0001928	<b>Repeated for each field</b>	<b>Repeated for each field</b>
<b>Easy bleeding</b> - Bleeding with little or no trauma	Bleeding with minor or no trauma; HP:0011889		
<b>Easy bruising</b> - Bruising with little or no trauma	Easy bruising; HP:0000978		
<b>Persistent bleeding</b> - Bleeding that continues for a long period of time after trauma; difficulties getting bleeding to stop once it has started	Persistent bleeding after trauma; HP:0001934		
<b>Extramedullary hematopoiesis</b> - A specific blood issue where blood cells form and develop outside of the bone marrow	Extramedullary hematopoiesis; HP:0001978		
<b>Frequent nosebleeds</b> - Frequent occurrence of nosebleeds (epistaxis)	Epistaxis; HP: 0000421		
<b>Platelets</b> - An issue with too many or too few platelets (thrombocytosis or thrombocytopenia)	Abnormality of thrombocytes; HP: 0001872		
<b>Red blood cells</b> - An issue with red blood cells, leading to problems such as anemia, polycythemia, or thalassemia, etc.	Abnormality of erythrocytes; HP:0001877		
<b>White blood cells</b> - An issue with too many or too few white blood cells (leukocytosis or leukopenia)	Abnormality of leukocytes; HP:0001881		

**Are there any other specific BLOOD/BLEEDING issues you've had that were not listed above? Please include at what age did the BLOOD/BLEEDING issues begin, and when were they diagnosed by a healthcare provider.**

## GenomeConnect Survey

What specific issues with your ORAL Health have you had? Please select all that apply.

Answers	HPO Term
<b>Gum disease</b> - Loss of tissue around teeth	Periodontitis; HP:0000704
<b>Gingivitis</b> - Early gum disease, inflamed gums, gingival inflammation, red and swollen gums	Gingival inflammation; HP:0000230
<b>Oral Herpes</b> - Blisters and ulcers on the gums and tongue caused by herpes virus	Oral herpes; HP:0410028
<b>Spontaneous tooth fracture</b> - A tendency of teeth to easily fracture or crack as manifested by a history of repeated fracture of the dental enamel – not caused by injury or dental restorations.	Spontaneous tooth fracture; HP:0025124
<b>Unusual number of teeth</b> - More/less teeth than normal	Abnormal number of teeth; HP:0006483
<b>Malformed teeth</b> - Misshapen teeth, abnormality of shape of tooth, deformity of teeth	Malformed teeth; HP:0006482
<b>Abnormality of primary teeth</b> - Baby teeth problems	Abnormality of primary teeth; HP:0006481
<b>Abnormality of the dental root</b> - Abnormal size or shape of the root(s) of teeth	Abnormality of the dental root; HP:0006486
<b>Abnormality of dental pulp</b> - Problems with the soft tissue within the pulp cavity located in the center of a tooth.	Abnormality of dental pulp; HP:0006479
<b>Over/under bite</b> - The teeth of the upper and lower jaw do not align well causing teeth of one jaw to be far in front of the other.	Misalalignment of teeth; HP:0000692
<b>None of the above</b>	
<b>Prefer not to answer</b>	

Are there any other specific ORAL HEALTH issues you've had that were not listed above?

# GenomeConnect Survey

## Reference

**Who referred you to the GenomeConnect site, or how did you find out about it? (Select all that apply.)**

Physician/doctor  
Genetic counselor  
Google search  
Other search engine (Yahoo, Bing, etc.)  
Laboratory report  
Social media (Facebook, Twitter, etc.)  
Patient support or advocacy group  
Friend  
Family member  
Unsure

**Are you already registered with or a member of a patient support, advocacy, or research group?**

- Yes
- No
- Unsure

**To which group do you belong?**

- None - I don't belong to a group
- Unsure
- Undiagnosed Disease Network (UDN)
- Unique
- Syndromes without a Name (SWAN, including all SWAN groups)
- Rare Genomics Institute
- Global Genes
- In Need of Diagnosis
- Genetic Alliance
- Syndrome-specific group (specify group name below)

**Are there additional thoughts you would like to share with us?**