

# GenomeConnect Survey

## GenomeConnect - Consent (Page 1)

**I understand the information about the purpose of this registry.**  
I have read and understood the information

## GenomeConnect - Consent (Page 2)

**I understand this information about the requirements for participation in GenomeConnect.**  
I have read and understood the information

### Who is being consented to participate.

Yourself  
Your child (ages newborn - 9 years)  
Your child (10 - 17 years old)  
Adult (age 18 or older for whom you have authority to consent)  
Unsure

## GenomeConnect - Consent (Page 3)

**I understand this section about who has access and what deidentified information is shared.**  
I have read and understood the information

## GenomeConnect - Consent (Page 4)

**I understand this information about the risks and benefits of participating in GenomeConnect.**  
I have read and understood the information

## GenomeConnect - Consent (Page 5)

**I understand that I can choose to stop participating in the registry at any time and have been told how to contact study staff for more information.**  
I have read and understood the information

## GenomeConnect - Assent

**I understand the contents of this form. I have had the opportunity to ask any questions, and if I had questions, they have been answered. I understand that I can print a copy of this form.**  
[Click to print a copy of the assent form.](#)  
I have read and understood the information

## GenomeConnect - Consent (Final Page)

**I understand the contents of this form. I have had the opportunity to ask any questions, and if I had questions, they have been answered. I understand that I can print a copy of this form.**  
[Click to print a copy of consent form.](#)  
I have read and understood the information

## Health Survey Overview

### 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)

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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

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## 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

## Mother's Pregnancy

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

#### Answers

**Amniotic fluid issue** - Too much fluid (polyhydramnios) or too little fluid (oligohydramnios) in the amniotic sac

**Decreased fetal movement** - Baby moved less than expected during pregnancy

**Delivery complication** - Can include breech birth, forceps or vacuum delivery, or umbilical

**Time of symptoms**  
First trimester  
Second trimester

**Time of diagnosis**  
First trimester  
Second trimester

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cord wrapped around the baby's neck

**Maternal health problem** - An issue with the mother's health during pregnancy; can include issues such as high blood pressure, diabetes, or CMV infection, etc.

**Placental issue** - Placenta was not formed, attached, or working properly

**Pregnancy exposure** - Mother had an exposure that could cause harm to the baby, like alcohol, cigarettes, street drugs, chemicals, or radiation, etc.

**Prematurity** - Baby was born early, before 37 weeks gestation

**Umbilical cord issue** - Cord was not formed properly or had an issue like a knot

Third trimester  
At delivery  
Unsure

Third trimester  
At delivery  
Unsure

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

**General overgrowth** - Significant and/or lifelong large size or overgrowth, often due to a syndrome

**Obesity** - Significant and/or lifelong issues with weight not specifically related to an overgrowth syndrome

**Overgrowth that is not symmetric** - One side of the body is larger or longer than the other side (hemihypertrophy)

**Short stature** - Height is much shorter than is typical for your family and/or the average height for someone of your same age and sex

**Undergrowth** - Significant problems growing as expected during childhood (failure to thrive)

**Growth hormone deficiency** - Insufficient production of growth hormone from pituitary gland

**Growth hormone excess** - Too much production of growth hormone from pituitary gland; can lead to acromegaly

**Tall stature** - Height much taller than is typical for your family and/or the average height from someone of your same age and sex

### Age symptoms first appeared

Unsure

Before birth

0 - 3 months old

4 - 7 months old

8 - 11 months old

1 year old

2 years old

3 years old

4 years old

5 years old

6 years old

7 years old

8 years old

9 years old

10 years old

11 years old

12 years old

13 years old

14 years old

15 years old

16 years old

17 years old

18 years old

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32 years old

33 years old

34 years old

35 years old

36 years old

37 years old

38 years old

39 years old

40 years old

41 years old

42 years old

43 years old

44 years old

45 years old

46 years old

47 years old

### Age at diagnosis

Unsure

Before birth

0 - 3 months old

4 - 7 months old

4 - 7 months old

8 - 11 months old

1 year old

2 years old

3 years old

4 years old

5 years old

6 years old

7 years old

8 years old

9 years old

10 years old

11 years old

12 years old

13 years old

14 years old

15 years old

16 years old

17 years old

18 years old

19 years old

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21 years old

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31 years old

32 years old

33 years old

34 years old

35 years old

36 years old

37 years old

38 years old

39 years old

40 years old

41 years old

42 years old

43 years old

44 years old

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48 years old	45 years old
49 years old	46 years old
50 years old	47 years old
51 years old	48 years old
52 years old	49 years old
53 years old	50 years old
54 years old	51 years old
55 years old	52 years old
56 years old	53 years old
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58 years old	55 years old
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63 years old	60 years old
64 years old	61 years old
65 years old	62 years old
66 years old	63 years old
67 years old	64 years old
68 years old	65 years old
69 years old	66 years old
70 and older	67 years old
	68 years old
	69 years old
	70 and older

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

**Type 2 diabetes** - The most common form of diabetes, sometimes called adult, late-onset, or non-insulin dependent; may or may not require insulin

**Type 1 diabetes** - Insulin-dependent diabetes, sometimes also called juvenile or autoimmune diabetes; typically requires insulin

**Delayed puberty** - Passing the age when puberty normally occurs without any signs of physical or hormonal puberty

**Diabetes insipidus** - Less-common form of diabetes showing extreme thirst and frequent urination

### Growth hormone deficiency

**Early puberty** - Puberty before the age of 8 years in girls or 9 years in boys; also called precocious puberty

**Hypogonadism** - Little or no sex hormones produced by the testes in men or the ovaries in women

**Adrenal hyperplasia** - Enlargement of the adrenal gland leading to decreased production of the hormone cortisol

**Goiter** - Enlargement of the thyroid gland which may result in neck swelling

**Hyperthyroidism** - High levels of thyroid hormone, including Graves' disease

**Hypothyroidism** - Low levels of thyroid hormone

**Parathyroid issue** - Too much or too little parathyroid hormone produced

### Age symptoms first appeared

Unsure

Before birth

0 - 3 months old

4 - 7 months old

8 - 11 months old

1 year old

2 years old

3 years old

4 years old

5 years old

6 years old

7 years old

8 years old

9 years old

10 years old

11 years old

12 years old

13 years old

14 years old

15 years old

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25 years old

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27 years old

28 years old

29 years old

30 years old

31 years old

32 years old

33 years old

34 years old

35 years old

36 years old

37 years old

38 years old

39 years old

### Age at diagnosis

Unsure

Before birth

0 - 3 months old

4 - 7 months old

8 - 11 months old

1 year old

2 years old

3 years old

4 years old

5 years old

6 years old

7 years old

8 years old

9 years old

10 years old

11 years old

12 years old

13 years old

14 years old

15 years old

16 years old

17 years old

18 years old

19 years old

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24 years old

25 years old

26 years old

27 years old

28 years old

29 years old

30 years old

31 years old

32 years old

33 years old

34 years old

35 years old

36 years old

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40 years old	37 years old
41 years old	38 years old
42 years old	39 years old
43 years old	40 years old
44 years old	41 years old
45 years old	42 years old
46 years old	43 years old
47 years old	44 years old
48 years old	45 years old
49 years old	46 years old
50 years old	47 years old
51 years old	48 years old
52 years old	49 years old
53 years old	50 years old
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64 years old	61 years old
65 years old	62 years old
66 years old	63 years old
67 years old	64 years old
68 years old	65 years old
69 years old	66 years old
70 and older	67 years old
	68 years old
	69 years old
	70 and older

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

What type of cancer was diagnosed? At what age did the cancer symptoms begin, and when was cancer diagnosed by a healthcare provider?

Answers	Age symptoms first appeared	Age at diagnosis
<b>Adrenocortical carcinoma</b> - A tumor in the adrenal gland which affects hormone production	Unsure	Unsure
<b>Breast cancer</b>	Before birth	Before birth
<b>Colon cancer</b>	0 - 3 months old	0 - 3 months old
<b>Hemangioblastoma</b> - A tumor of blood vessel cells originating from the central nervous system	4 - 7 months old	old
<b>Intestinal tumors or polyps</b> - Multiple polyps or tumors in the intestines, not just one or a few; sometimes called polyposis	8 - 11 months old	4 - 7 months old
<b>Kidney cancer</b>	1 year old	old
<b>Leukemia, juvenile myelomonocytic</b> - JML	2 years old	8 - 11 months old
<b>Leukemia, no specific type</b> - Any type of leukemia that is NOT JML	3 years old	old
<b>Lung cancer</b>	4 years old	1 year old
<b>Lymphoma</b> - Blood cell tumors developing from white blood cells; can include non-Hodgkin, Hodgkin, multiple myeloma, or immunoproliferative disease	5 years old	2 years old
<b>Malignant nerve sheath tumor</b> - Also called MPNST, malignant schwannoma, or neurofibrosarcoma; a tumor of connective tissue surrounding nerves	6 years old	3 years old
<b>Medullary thyroid carcinoma</b> - A neuroendocrine tumor in the thyroid gland	7 years old	4 years old
<b>Mouth cancer</b> - Includes oral cancer and cancer of the mouth but NOT throat cancer	8 years old	5 years old
<b>Osteosarcoma</b>	9 years old	6 years old
<b>Ovarian cancer</b>	10 years old	7 years old
<b>Pancreatic cancer</b>	11 years old	8 years old
<b>Paraganglioma</b> - A tumor of neuroendocrine tissue at the base of skull, in the neck, or along the spine	12 years old	9 years old
<b>Pheochromocytoma</b> - A specific type of paraganglioma located in the adrenal gland	13 years old	10 years old
<b>Pleuropulmonary blastoma</b> - A type of lung tumor more commonly affecting infants/children	14 years old	11 years old
<b>Prostate cancer</b>	15 years old	12 years old
<b>Testicular cancer</b>	16 years old	13 years old
<b>Retinoblastoma</b> - A tumor of retinal cells in the eye	17 years old	14 years old
<b>Schwannoma</b> - Also called neuroma or Schwann cell tumor, a type of slow-growing and typically benign nerve sheath tumor	18 years old	15 years old
<b>Skin cancer (melanoma)</b>	19 years old	16 years old
<b>Skin cancer (non-melanoma)</b> - Any skin cancer that is NOT melanoma	20 years old	17 years old
<b>Stomach cancer</b>	21 years old	18 years old
<b>Uterine cancer</b> - Also called endometrial cancer	22 years old	19 years old
<b>Have had cancer</b> -but not sure what type	23 years old	20 years old
	24 years old	21 years old
	25 years old	22 years old
	26 years old	23 years old
	27 years old	24 years old
	28 years old	25 years old
	29 years old	26 years old
	30 years old	27 years old
	31 years old	28 years old

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**Have had cancer** - but type not on this list - Please specify your cancer name/type below

- |              |              |
|--------------|--------------|
| 32 years old | 29 years old |
| 33 years old | 30 years old |
| 34 years old | 31 years old |
| 35 years old | 32 years old |
| 36 years old | 33 years old |
| 37 years old | 34 years old |
| 38 years old | 35 years old |
| 39 years old | 36 years old |
| 40 years old | 37 years old |
| 41 years old | 38 years old |
| 42 years old | 39 years old |
| 43 years old | 40 years old |
| 44 years old | 41 years old |
| 45 years old | 42 years old |
| 46 years old | 43 years old |
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| 65 years old | 62 years old |
| 66 years old | 63 years old |
| 67 years old | 64 years old |
| 68 years old | 65 years old |
| 69 years old | 66 years old |
| 70 and older | 67 years old |
|              | 68 years old |
|              | 69 years old |
|              | 70 and older |

**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

**Chin** - Can include differences in shape or structure of chin, such as an underdeveloped or recessed appearance, etc.

**Cleft lip and/or cleft palate** - A "split" or division in the lip and/or roof of the mouth present at birth

**General face shape** - Facial features may have been described as atypical or have a shape/look that differs from the rest of the family

**Hair** - Can include having too little or "sparse" hair or a hair texture different from what is expected, etc.

**Inside of the mouth** - Can include issues with inside of the mouth, teeth, gums, or tongue

**Lips or outside of the mouth** - Can include issues with lip or mouth size, shape, or overall development

**Neck** - Can include a short neck, broad neck, webbed neck, branchial clefts, branchial pits, or branchial cysts, etc.

**Nose** - Broad category that can include a significant problem with the way the nose was formed or the way that it works

**Skull** - 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.

**Throat/swallowing difficulties** - Also called oral pharyngeal dysphagia

### Age symptoms first appeared

- Unsure
- Before birth
- 0 - 3 months old
- 4 - 7 months old
- 8 - 11 months old
- 1 year old
- 2 years old
- 3 years old
- 4 years old
- 5 years old
- 6 years old
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- 16 years old
- 17 years old
- 18 years old
- 19 years old
- 20 years old

### Age at diagnosis

- Unsure
- Before birth
- 0 - 3 months old
- 4 - 7 months old
- 8 - 11 months old
- 1 year old
- 2 years old
- 3 years old
- 4 years old
- 5 years old
- 6 years old
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- 15 years old
- 16 years old
- 17 years old

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21 years old	18 years old
22 years old	19 years old
23 years old	20 years old
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25 years old	22 years old
26 years old	23 years old
27 years old	24 years old
28 years old	25 years old
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65 years old	62 years old
66 years old	63 years old
67 years old	64 years old
68 years old	65 years old
69 years old	66 years old
70 and older	67 years old
	68 years old
	69 years old
	70 and older

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?

### Answers

**Eye movement issues** - Eyes have unusual movement such as "lazy eye," (strabismus) or eye tremors (nystagmus), etc.

**Eye size difference** - Eye size differs from what is usual such as a small eye size (microphthalmia) or lack of eye development (anophthalmia), etc.

**Iris/pupil difference** - The colored part of the eye, called the iris, has freckles/spots, eye color differs between eyes, or the pupil has notches (coloboma), etc.

**Lens issue** - A cataract or dislocated lens (ectopia lentis), etc.

**Major vision issues** - Issues may include blindness, color blindness, night vision problems, floaters, or light sensitivity, etc.; do NOT include minor nearsightedness or farsightedness.

**Nearsightedness** - Also called myopia; things in the distance look fuzzy

**Farsightedness** - Also called hyperopia or hypermetropia; things up close look fuzzy

**Optic nerve issues** - 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

**Retina issues** - 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 degeneration, or retinitis pigmentosa, etc.

### Age symptoms first appeared

Unsure  
Before birth  
0 - 3 months old  
4 - 7 months old  
8 - 11 months old  
1 year old  
2 years old  
3 years old  
4 years old  
5 years old  
6 years old  
7 years old  
8 years old  
9 years old  
10 years old  
11 years old  
12 years old

### Age at diagnosis

Unsure  
Before birth  
0 - 3 months old  
4 - 7 months old  
8 - 11 months old  
8 - 11 months old  
1 year old  
2 years old  
3 years old  
4 years old  
5 years old  
6 years old  
7 years old  
8 years old  
9 years old



# GenomeConnect Survey

**Ptosis** - Droopiness of the eyelid

- |              |              |
|--------------|--------------|
| 13 years old | 10 years old |
| 14 years old | 11 years old |
| 15 years old | 12 years old |
| 16 years old | 13 years old |
| 17 years old | 14 years old |
| 18 years old | 15 years old |
| 19 years old | 16 years old |
| 20 years old | 17 years old |
| 21 years old | 18 years old |
| 22 years old | 19 years old |
| 23 years old | 20 years old |
| 24 years old | 21 years old |
| 25 years old | 22 years old |
| 26 years old | 23 years old |
| 27 years old | 24 years old |
| 28 years old | 25 years old |
| 29 years old | 26 years old |
| 30 years old | 27 years old |
| 31 years old | 28 years old |
| 32 years old | 29 years old |
| 33 years old | 30 years old |
| 34 years old | 31 years old |
| 35 years old | 32 years old |
| 36 years old | 33 years old |
| 37 years old | 34 years old |
| 38 years old | 35 years old |
| 39 years old | 36 years old |
| 40 years old | 37 years old |
| 41 years old | 38 years old |
| 42 years old | 39 years old |
| 43 years old | 40 years old |
| 44 years old | 41 years old |
| 45 years old | 42 years old |
| 46 years old | 43 years old |
| 47 years old | 44 years old |
| 48 years old | 45 years old |
| 49 years old | 46 years old |
| 50 years old | 47 years old |
| 51 years old | 48 years old |
| 52 years old | 49 years old |
| 53 years old | 50 years old |
| 54 years old | 51 years old |
| 55 years old | 52 years old |
| 56 years old | 53 years old |
| 57 years old | 54 years old |
| 58 years old | 55 years old |
| 59 years old | 56 years old |
| 60 years old | 57 years old |
| 61 years old | 58 years old |
| 62 years old | 59 years old |
| 63 years old | 60 years old |
| 64 years old | 61 years old |
| 65 years old | 62 years old |
| 66 years old | 63 years old |
| 67 years old | 64 years old |
| 68 years old | 65 years old |
| 69 years old | 66 years old |
| 70 and older | 67 years old |
|              | 68 years old |
|              | 69 years old |
|              | 70 and older |

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

**Ear shape difference** - 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.

**Conductive hearing loss** - Hearing loss due to an issue with the ear canal, ear drum, or the bones in the middle ear

**Sensorineural hearing loss (SNHL)** - Hearing loss due to an issue with the nerves in the inner ear or connecting ear to the brain

**Mixed hearing loss** - A combination of both conductive and sensorineural hearing loss

**Hearing loss, unsure which type**

Age symptoms first appeared	Age at diagnosis
Unsure	Unsure
Before birth	Before birth
0 - 3 months old	0 - 3 months old
4 - 7 months old	old
8 - 11 months old	4 - 7 months old
1 year old	old
2 years old	8 - 11 months old
3 years old	months old
4 years old	1 year old

# GenomeConnect Survey

**Ringing in the ears** - Also called tinnitus

**Hyperacusis** - Oversensitivity to or difficulty tolerating certain sounds

**Dizziness** - Also called vertigo

5 years old	2 years old
6 years old	3 years old
7 years old	4 years old
8 years old	5 years old
9 years old	6 years old
10 years old	7 years old
11 years old	8 years old
12 years old	9 years old
13 years old	10 years old
14 years old	11 years old
15 years old	12 years old
16 years old	13 years old
17 years old	14 years old
18 years old	15 years old
19 years old	16 years old
20 years old	17 years old
21 years old	18 years old
22 years old	19 years old
23 years old	20 years old
24 years old	21 years old
25 years old	22 years old
26 years old	23 years old
27 years old	24 years old
28 years old	25 years old
29 years old	26 years old
30 years old	27 years old
31 years old	28 years old
32 years old	29 years old
33 years old	30 years old
34 years old	31 years old
35 years old	32 years old
36 years old	33 years old
37 years old	34 years old
38 years old	35 years old
39 years old	36 years old
40 years old	37 years old
41 years old	38 years old
42 years old	39 years old
43 years old	40 years old
44 years old	41 years old
45 years old	42 years old
46 years old	43 years old
47 years old	44 years old
48 years old	45 years old
49 years old	46 years old
50 years old	47 years old
51 years old	48 years old
52 years old	49 years old
53 years old	50 years old
54 years old	51 years old
55 years old	52 years old
56 years old	53 years old
57 years old	54 years old
58 years old	55 years old
59 years old	56 years old
60 years old	57 years old
61 years old	58 years old
62 years old	59 years old
63 years old	60 years old
64 years old	61 years old
65 years old	62 years old
66 years old	63 years old
67 years old	64 years old
68 years old	65 years old
69 years old	66 years old
70 and older	67 years old
	68 years old
	69 years old
	70 and older

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**

**Brain and/or spinal cord issue** - Includes problems with the formation or appearance of

**Age symptoms first appeared**  
 Unsure

**Age at diagnosis**  
 Unsure

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the brain and/or spinal cord, often found by MRI

**Cerebral palsy (CP)** - A single damaging event to the brain that can cause movement problems, seizures, learning problems, and/or communication problems

**Cognitive impairment** - Significant difficulties learning, understanding, or processing information, or making decisions

**Coordination issues** - Significant clumsiness or other difficulties with movement due to body parts not working together as they normally would

**EEG abnormality** - A problem has been identified on an electroencephalogram, or "EEG," a test that measures the electrical activity of the brain

**Encephalopathy** - Damage to the brain, sometimes ongoing, that results in altered brain function

**Hypertonia** - Muscle tightness that limits movement, spasticity, increased muscle tone

**Hypotonia** - Unusually low muscle tone, muscle tone may have been described as floppy

**Memory impairment** - Increased forgetfulness or significant difficulty remembering things

**Unusual movements** - Changes or problems in the ability to move body parts voluntarily; do NOT include Parkinson's Disease here

**Parkinson's disease** - Abnormal movements, characterized by shaking, stiffness, slowness, and difficulty walking

**Seizures** - Also known as convulsions

Before birth

0 - 3 months old

4 - 7 months old

8 - 11 months old

1 year old

2 years old

3 years old

4 years old

5 years old

6 years old

7 years old

8 years old

9 years old

10 years old

11 years old

12 years old

13 years old

14 years old

15 years old

16 years old

17 years old

18 years old

19 years old

20 years old

21 years old

22 years old

23 years old

24 years old

25 years old

26 years old

27 years old

28 years old

29 years old

30 years old

31 years old

32 years old

33 years old

34 years old

35 years old

36 years old

37 years old

38 years old

39 years old

40 years old

41 years old

42 years old

43 years old

44 years old

45 years old

46 years old

47 years old

48 years old

49 years old

50 years old

51 years old

52 years old

53 years old

54 years old

55 years old

56 years old

57 years old

58 years old

59 years old

60 years old

61 years old

62 years old

63 years old

64 years old

65 years old

66 years old

67 years old

68 years old

69 years old

70 and older

Before birth

0 - 3 months old

4 - 7 months old

8 - 11 months old

1 year old

2 years old

3 years old

4 years old

5 years old

6 years old

7 years old

8 years old

9 years old

10 years old

11 years old

12 years old

13 years old

14 years old

15 years old

16 years old

17 years old

18 years old

19 years old

20 years old

21 years old

22 years old

23 years old

24 years old

25 years old

26 years old

27 years old

28 years old

29 years old

30 years old

31 years old

32 years old

33 years old

34 years old

35 years old

36 years old

37 years old

38 years old

39 years old

40 years old

41 years old

42 years old

43 years old

44 years old

45 years old

46 years old

47 years old

48 years old

49 years old

50 years old

51 years old

52 years old

53 years old

54 years old

55 years old

56 years old

57 years old

58 years old

59 years old

60 years old

61 years old

62 years old

63 years old

64 years old

65 years old

66 years old

67 years old

68 years old

69 years old

70 and older

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

# GenomeConnect Survey

## 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

**Autism spectrum disorder (ASD)** - Includes autism and pervasive developmental disorder, not otherwise specified (PDD-NOS)

**Bipolar disorder** - Also known as manic depression; characterized by severe mood swings that affect the ability to perform day-to-day activities

**Schizophrenia** - A severe disorder in which people have difficulty determining what is real and unreal, and have unusual social behaviors

**Repetitive or self-injurious behavior** - Can include issues such as body-rocking, head-banging, hand-wringing, or skin picking, etc.

### Aggressive/violent behavior

**Anxiety** - Feelings of dread or fear that interfere with daily living

### Depression

**Delusions** - Believing that something is true despite evidence showing it is not

**Hallucinations** - Seeing or hearing things that are not real while you are awake

**Obsessive compulsive disorder (OCD)** - The need to perform recurrent rituals that interfere with your daily life

### Psychosis

**Short attention span** - Includes attention deficit disorder (ADD) and attention deficit hyperactivity disorder (ADHD)

Age symptoms first appeared	Age at diagnosis
Unsure	Unsure
Before birth	Before birth
0 - 3 months old	0 - 3 months old
4 - 7 months old	4 - 7 months old
8 - 11 months old	8 - 11 months old
1 year old	1 year old
2 years old	2 years old
3 years old	3 years old
4 years old	4 years old
5 years old	5 years old
6 years old	6 years old
7 years old	7 years old
8 years old	8 years old
9 years old	9 years old
10 years old	10 years old
11 years old	11 years old
12 years old	12 years old
13 years old	13 years old
14 years old	14 years old
15 years old	15 years old
16 years old	16 years old
17 years old	17 years old
18 years old	18 years old
19 years old	19 years old
20 years old	20 years old
21 years old	21 years old
22 years old	22 years old
23 years old	23 years old
24 years old	24 years old
25 years old	25 years old
26 years old	26 years old
27 years old	27 years old
28 years old	28 years old
29 years old	29 years old
30 years old	30 years old
31 years old	31 years old
32 years old	32 years old
33 years old	33 years old
34 years old	34 years old
35 years old	35 years old
36 years old	36 years old
37 years old	37 years old
38 years old	38 years old
39 years old	39 years old
40 years old	40 years old
41 years old	41 years old
42 years old	42 years old
43 years old	43 years old
44 years old	44 years old
45 years old	45 years old
46 years old	46 years old
47 years old	47 years old
48 years old	48 years old
49 years old	49 years old
50 years old	50 years old
51 years old	51 years old
52 years old	52 years old
53 years old	53 years old
54 years old	54 years old
55 years old	55 years old
56 years old	56 years old
57 years old	57 years old
58 years old	58 years old
59 years old	59 years old
60 years old	60 years old
61 years old	61 years old
62 years old	62 years old
63 years old	63 years old
64 years old	64 years old
65 years old	65 years old
66 years old	66 years old
67 years old	67 years old
68 years old	68 years old

# GenomeConnect Survey

69 years old  
70 and older

66 years old  
67 years old  
68 years old  
69 years old  
70 and older

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

**Atrophic scars** - Scars that retract or indent

**"Café-au-lait" spots** - Light brown birth marks the color of coffee with milk

**Hyperpigmentation** - Patches of skin that are darker than the regular skin color

**Hypopigmentation** - Patches of skin that are LIGHTER than the regular skin color

**Decreased sweating** - Also called hypohidrosis; you are not able to sweat or your ability to sweat is significantly reduced

**Increased sweating** - Also called "hyperhidrosis"; your sweating is significantly increased, above what is necessary to maintain a normal body temperature

**Skin fragility or sensitivity** - Skin is fragile to touch or sunlight; can include issues like ectodermal dysplasia, premature aging of the skin, or extreme light sensitivity, etc.

**Thick skin** - 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

**Hyperelastic skin** - Skin that is extremely stretchy and returns to its original position after stretching

**Hypoelastic skin** - Inelastic or "saggy" skin, often hanging in loose folds (cutis laxa)

### Age symptoms first appeared

Unsure  
Before birth  
0 - 3 months old  
4 - 7 months old  
8 - 11 months old  
1 year old  
2 years old  
3 years old  
4 years old  
5 years old  
6 years old  
7 years old  
8 years old  
9 years old  
10 years old  
11 years old  
12 years old  
13 years old  
14 years old  
15 years old  
16 years old  
17 years old  
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22 years old  
23 years old  
24 years old  
25 years old  
26 years old  
27 years old  
28 years old  
29 years old  
30 years old  
31 years old  
32 years old  
33 years old  
34 years old  
35 years old  
36 years old  
37 years old  
38 years old  
39 years old  
40 years old  
41 years old  
42 years old  
43 years old  
44 years old  
45 years old  
46 years old  
47 years old  
48 years old  
49 years old  
50 years old  
51 years old  
52 years old  
53 years old  
54 years old  
55 years old  
56 years old  
57 years old  
58 years old  
59 years old  
60 years old

### Age at diagnosis

Unsure  
Before birth  
0 - 3 months old  
4 - 7 months old  
8 - 11 months old  
1 year old  
2 years old  
3 years old  
4 years old  
5 years old  
6 years old  
7 years old  
8 years old  
9 years old  
10 years old  
11 years old  
12 years old  
13 years old  
14 years old  
15 years old  
16 years old  
17 years old  
18 years old  
19 years old  
20 years old  
21 years old  
22 years old  
23 years old  
24 years old  
25 years old  
26 years old  
27 years old  
28 years old  
29 years old  
30 years old  
31 years old  
32 years old  
33 years old  
34 years old  
35 years old  
36 years old  
37 years old  
38 years old  
39 years old  
40 years old  
41 years old  
42 years old  
43 years old  
44 years old  
45 years old  
46 years old  
47 years old  
48 years old  
49 years old  
50 years old  
51 years old  
52 years old  
53 years old  
54 years old  
55 years old  
56 years old  
57 years old

# GenomeConnect Survey

61 years old	58 years old
62 years old	59 years old
63 years old	60 years old
64 years old	61 years old
65 years old	62 years old
66 years old	63 years old
67 years old	64 years old
68 years old	65 years old
69 years old	66 years old
70 and older	67 years old
	68 years old
	69 years old
	70 and older

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?

### Answers

**Arm and/or leg bone differences since birth** - 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.

**Curvature of the spine** - Spine curves side-to-side (scoliosis) or front-to-back (kyphosis)

**Flexible joints** - Commonly called being double-jointed, hypermobile, or hyper extensible

**Frequent broken bones** - Bones tend to break with minimal trauma, includes "brittle bone disease"

**Hand, foot, finger, and/or toe differences** - Includes bony differences such as a "split" hand, extra, missing, or fused fingers and/or toes, etc.

**Hip dysplasia** - Issues with the formation of the hip socket, typically present at birth

**Pectus carinatum** - Breastbone sticks out from the chest, sometimes called "pigeon chest"

**Pectus excavatum** - Breastbone appears sunken or caved in to the chest, sometimes called "funnel chest"

**Skeletal dysplasia** - Issues or differences with the growth of the bones and/or cartilage, sometimes called dwarfism

Age symptoms first appeared	Age at diagnosis
Unsure	Unsure
Before birth	Before birth
0 - 3 months old	0 - 3 months old
4 - 7 months old	4 - 7 months old
8 - 11 months old	8 - 11 months old
1 year old	1 year old
2 years old	2 years old
3 years old	3 years old
4 years old	4 years old
5 years old	5 years old
6 years old	6 years old
7 years old	7 years old
8 years old	8 years old
9 years old	9 years old
10 years old	10 years old
11 years old	11 years old
12 years old	12 years old
13 years old	13 years old
14 years old	14 years old
15 years old	15 years old
16 years old	16 years old
17 years old	17 years old
18 years old	18 years old
19 years old	19 years old
20 years old	20 years old
21 years old	21 years old
22 years old	22 years old
23 years old	23 years old
24 years old	24 years old
25 years old	25 years old
26 years old	26 years old
27 years old	27 years old
28 years old	28 years old
29 years old	29 years old
30 years old	30 years old
31 years old	31 years old
32 years old	32 years old
33 years old	33 years old
34 years old	34 years old
35 years old	35 years old
36 years old	36 years old
37 years old	37 years old
38 years old	38 years old
39 years old	39 years old
40 years old	40 years old
41 years old	41 years old
42 years old	42 years old
43 years old	43 years old
44 years old	44 years old
45 years old	45 years old
46 years old	46 years old
47 years old	47 years old
48 years old	48 years old
49 years old	49 years old
50 years old	50 years old
51 years old	51 years old
52 years old	52 years old

# GenomeConnect Survey

53 years old	50 years old
54 years old	51 years old
55 years old	52 years old
56 years old	53 years old
57 years old	54 years old
58 years old	55 years old
59 years old	56 years old
60 years old	57 years old
61 years old	58 years old
62 years old	59 years old
63 years old	60 years old
64 years old	61 years old
65 years old	62 years old
66 years old	63 years old
67 years old	64 years old
68 years old	65 years old
69 years old	66 years old
70 and older	67 years old
	68 years old
	69 years old
	70 and older

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

**Facial muscle issue** - Issues with the muscles of the face; can include facial palsy, difficulty with chewing, or problems moving your eyes, etc.

**Issue with muscle function** - Issues with the way the muscles WORK/FEEL; can include weakness, stiffness, pain, involuntary movements, cramping, or low muscle tone, etc.

**Issue with internal muscle structure** - Issues with the way the muscles are formed; often diagnosed by a muscle biopsy

**Issue with nerves connected to muscles** - Issues with the way nerves communicate with muscles; often diagnosed with nerve conduction studies or electromyography (EMG)

**Muscle issues in the arms and/or legs**- Weakness or underdevelopment

**Muscle issues in the chest, back, and/or shoulders** - Weakness or underdevelopment

**Neck muscle issue** - Torticollis or neck weakness

Age symptoms first appeared	Age at diagnosis
Unsure	Unsure
Before birth	Before birth
0 - 3 months old	0 - 3 months old
4 - 7 months old	4 - 7 months old
8 - 11 months old	8 - 11 months old
1 year old	1 year old
2 years old	2 years old
3 years old	3 years old
4 years old	4 years old
5 years old	5 years old
6 years old	6 years old
7 years old	7 years old
8 years old	8 years old
9 years old	9 years old
10 years old	10 years old
11 years old	11 years old
12 years old	12 years old
13 years old	13 years old
14 years old	14 years old
15 years old	15 years old
16 years old	16 years old
17 years old	17 years old
18 years old	18 years old
19 years old	19 years old
20 years old	20 years old
21 years old	21 years old
22 years old	22 years old
23 years old	23 years old
24 years old	24 years old
25 years old	25 years old
26 years old	26 years old
27 years old	27 years old
28 years old	28 years old
29 years old	29 years old
30 years old	30 years old
31 years old	31 years old
32 years old	32 years old
33 years old	33 years old
34 years old	34 years old
35 years old	35 years old
36 years old	36 years old
37 years old	37 years old
38 years old	38 years old
39 years old	39 years old
40 years old	40 years old
41 years old	41 years old
42 years old	42 years old
43 years old	43 years old

# GenomeConnect Survey

44 years old	41 years old
45 years old	42 years old
46 years old	43 years old
47 years old	44 years old
48 years old	45 years old
49 years old	46 years old
50 years old	47 years old
51 years old	48 years old
52 years old	49 years old
53 years old	50 years old
54 years old	51 years old
55 years old	52 years old
56 years old	53 years old
57 years old	54 years old
58 years old	55 years old
59 years old	56 years old
60 years old	57 years old
61 years old	58 years old
62 years old	59 years old
63 years old	60 years old
64 years old	61 years old
65 years old	62 years old
66 years old	63 years old
67 years old	64 years old
68 years old	65 years old
69 years old	66 years old
70 and older	67 years old
	68 years old
	69 years old
	70 and older

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

- Aneurysm** - An enlarged or bulging blood vessel
- Arrhythmia** - A heart rhythm that is too fast, slow, or irregular
- Abnormal EKG (ECG)** - Issues with the electrical signals of the heart, seen on electrocardiogram
- Arterial dissection** - A tear or rip in a large blood vessel
- Cardiomyopathy** - Issues with the heart muscle; can include enlargement, thickening, or scarring
- Fainting** - Passing out (syncope)
- Heart defect** - Issues with the way the heart and/or vessels were formed, often diagnosed at birth
- High blood pressure** - Also called hypertension; if this is known to be related to another condition you have, like pregnancy or pheochromocytoma, please provide more information below
- High cholesterol** - High levels of cholesterol in the blood, also called hypercholesterolemia
- Stroke** - Loss of blood supply to the brain

Age symptoms first appeared	Age at diagnosis
Unsure	Unsure
Before birth	Before birth
0 - 3 months old	0 - 3 months old
4 - 7 months old	old
8 - 11 months old	4 - 7 months old
1 year old	8 - 11 months old
2 years old	1 year old
3 years old	2 years old
4 years old	3 years old
5 years old	4 years old
6 years old	5 years old
7 years old	6 years old
8 years old	7 years old
9 years old	8 years old
10 years old	9 years old
11 years old	10 years old
12 years old	11 years old
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18 years old	17 years old
19 years old	18 years old
20 years old	19 years old
21 years old	20 years old
22 years old	21 years old
23 years old	22 years old
24 years old	23 years old
25 years old	24 years old
26 years old	25 years old
27 years old	26 years old
28 years old	27 years old
29 years old	28 years old
30 years old	29 years old
31 years old	30 years old
32 years old	31 years old
33 years old	32 years old
34 years old	
35 years old	



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36 years old	33 years old
37 years old	34 years old
38 years old	35 years old
39 years old	36 years old
40 years old	37 years old
41 years old	38 years old
42 years old	39 years old
43 years old	40 years old
44 years old	41 years old
45 years old	42 years old
46 years old	43 years old
47 years old	44 years old
48 years old	45 years old
49 years old	46 years old
50 years old	47 years old
51 years old	48 years old
52 years old	49 years old
53 years old	50 years old
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61 years old	58 years old
62 years old	59 years old
63 years old	60 years old
64 years old	61 years old
65 years old	62 years old
66 years old	63 years old
67 years old	64 years old
68 years old	65 years old
69 years old	66 years old
70 and older	67 years old
	68 years old
	69 years old
	70 and older

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

#### Asthma

**Chronic lung issues** - 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

**Decreased lung function** - Issues with lungs working at "full capacity"; also called decreased pulmonary function

**Diaphragm issues** - Issues with the muscle that separates your chest from your abdomen such as congenital diaphragmatic hernia or weakness of the diaphragm

**Respiratory insufficiency** - Issues with taking in enough oxygen and/or getting rid of enough carbon dioxide

**Restrictive lung disease** - Issues with fully expanding the lungs with air; can be caused by issues outside of the lungs such as neuromuscular disorders, scoliosis, or sarcoidosis

**Unusual breathing patterns** - Issues include apnea, hyperventilation, or rapid breathing (tachypnea)

**Upper respiratory tract issues** - Issues with the voice box (larynx), windpipe (trachea), or air tubes leading to lungs (bronchi)

### Age symptoms first appeared

Unsure  
Before birth  
0 - 3 months old  
4 - 7 months old  
8 - 11 months old  
1 year old  
2 years old  
3 years old  
4 years old  
5 years old  
6 years old  
7 years old  
8 years old  
9 years old  
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20 years old  
21 years old  
22 years old  
23 years old  
24 years old  
25 years old  
26 years old  
27 years old

### Age at diagnosis

Unsure  
Before birth  
0 - 3 months old  
4 - 7 months old  
8 - 11 months old  
1 year old  
2 years old  
3 years old  
4 years old  
5 years old  
6 years old  
7 years old  
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15 years old  
16 years old  
17 years old  
18 years old  
19 years old  
20 years old  
21 years old  
22 years old  
23 years old  
24 years old

# GenomeConnect Survey

28 years old	25 years old
29 years old	26 years old
30 years old	27 years old
31 years old	28 years old
32 years old	29 years old
33 years old	30 years old
34 years old	31 years old
35 years old	32 years old
36 years old	33 years old
37 years old	34 years old
38 years old	35 years old
39 years old	36 years old
40 years old	37 years old
41 years old	38 years old
42 years old	39 years old
43 years old	40 years old
44 years old	41 years old
45 years old	42 years old
46 years old	43 years old
47 years old	44 years old
48 years old	45 years old
49 years old	46 years old
50 years old	47 years old
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63 years old	60 years old
64 years old	61 years old
65 years old	62 years old
66 years old	63 years old
67 years old	64 years old
68 years old	65 years old
69 years old	66 years old
70 and older	67 years old
	68 years old
	69 years old
	70 and older

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

**Feeding difficulties** - Issues with eating; can include problems eating food by mouth or keeping food from being vomited

**Esophagus** - Issues with the tube that takes food from your mouth to your stomach; can include reflux, inflammation, or narrowing of the esophagus, etc.

**Stomach** - Issues with the stomach; can include a hernia, pyloric stenosis, or problems emptying food from the stomach, etc.

**Pancreas** - Issues with pancreas such as pancreatitis or cysts

**Liver** - Issues with liver; can include enlargement (hepatomegaly), scarring (cirrhosis), hepatitis, or cysts, etc.

**Dysmotility** - 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

**Intestines** - Issues with the small intestine, large intestine, bowel, and colon; can include malabsorption, intestinal malrotation, polyps, diverticulitis, Crohn's disease, or colitis, etc.

**Soiling/defecation** - Issues with producing a normal bowel movement (feces/poop); can include bowel incontinence, colitis, malrotation of the colon, etc.

**Anus** - 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

### Age symptoms first appeared

Unsure  
 Before birth  
 0 - 3 months old  
 4 - 7 months old  
 8 - 11 months old  
 1 year old  
 2 years old  
 3 years old  
 4 years old  
 5 years old  
 6 years old  
 7 years old  
 8 years old  
 9 years old  
 10 years old  
 11 years old  
 12 years old  
 13 years old  
 14 years old  
 15 years old  
 16 years old  
 17 years old  
 18 years old  
 19 years old

### Age at diagnosis

Unsure  
 Before birth  
 0 - 3 months old  
 4 - 7 months old  
 8 - 11 months old  
 1 year old  
 2 years old  
 3 years old  
 4 years old  
 5 years old  
 6 years old  
 7 years old  
 8 years old  
 9 years old  
 10 years old  
 11 years old  
 12 years old  
 13 years old  
 14 years old  
 15 years old  
 16 years old

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20 years old	17 years old
21 years old	18 years old
22 years old	19 years old
23 years old	20 years old
24 years old	21 years old
25 years old	22 years old
26 years old	23 years old
27 years old	24 years old
28 years old	25 years old
29 years old	26 years old
30 years old	27 years old
31 years old	28 years old
32 years old	29 years old
33 years old	30 years old
34 years old	31 years old
35 years old	32 years old
36 years old	33 years old
37 years old	34 years old
38 years old	35 years old
39 years old	36 years old
40 years old	37 years old
41 years old	38 years old
42 years old	39 years old
43 years old	40 years old
44 years old	41 years old
45 years old	42 years old
46 years old	43 years old
47 years old	44 years old
48 years old	45 years old
49 years old	46 years old
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63 years old	60 years old
64 years old	61 years old
65 years old	62 years old
66 years old	63 years old
67 years old	64 years old
68 years old	65 years old
69 years old	66 years old
70 and older	67 years old
	68 years old
	69 years old
	70 and older

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

**Bladder** - Issues with how the bladder is shaped or works; can include problems with bladder stones, bladder size, overactive bladder, or incontinence, etc.

**Internal or external sex organ disorder** - 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

**Kidney function issue** - Issue with how the kidneys work such as kidney failure or problems with proper filtering/absorption

**Kidney structure issue** - Issue with the kidney shape/structure; can include abnormal size, missing kidney, or abnormal kidney location in the body

**Female sex organ disorder** - Issues with the formation or appearance of the female genitalia, including labia, vagina, uterus, and/or ovaries

**Genitalia function** - Issues with the way the male or female genitalia works; can include infertility, impotence, or ovarian failure, etc.

**Male sex organ disorder** - Issues with the formation or appearance of the male genitalia, including penis, scrotum, testicles, and/or prostate gland

**Urethra issue** - Issues with the tube that empties the bladder; can include problems with way

### Age symptoms first appeared

Unsure  
Before birth  
0 - 3 months old  
4 - 7 months old  
8 - 11 months old  
1 year old  
2 years old  
3 years old  
4 years old  
5 years old  
6 years old  
7 years old  
8 years old  
9 years old  
10 years old

### Age at diagnosis

Unsure  
Before birth  
0 - 3 months old  
4 - 7 months old  
8 - 11 months old  
1 year old  
2 years old  
3 years old  
4 years old  
5 years old  
6 years old  
7 years old  
8 years old

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urethra formed, blockage, or abnormal urethra connection (hypospadias), etc.

**Ureter issue** - 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.

**Urine ("pee") issues** - Issues with urine; can include blood or protein in urine (hematuria or proteinuria), unusual urine color, or unusual urine odor, etc.

11 years old	9 years old
12 years old	10 years old
13 years old	11 years old
14 years old	12 years old
15 years old	13 years old
16 years old	14 years old
17 years old	15 years old
18 years old	16 years old
19 years old	17 years old
20 years old	18 years old
21 years old	19 years old
22 years old	20 years old
23 years old	21 years old
24 years old	22 years old
25 years old	23 years old
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37 years old	35 years old
38 years old	36 years old
39 years old	37 years old
40 years old	38 years old
41 years old	39 years old
42 years old	40 years old
43 years old	41 years old
44 years old	42 years old
45 years old	43 years old
46 years old	44 years old
47 years old	45 years old
48 years old	46 years old
49 years old	47 years old
50 years old	48 years old
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62 years old	60 years old
63 years old	61 years old
64 years old	62 years old
65 years old	63 years old
66 years old	64 years old
67 years old	65 years old
68 years old	66 years old
69 years old	67 years old
70 and older	68 years old
	69 years old
	70 and older

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

**Autoimmune disorder** - Disorder when the immune system attacks the body's own healthy tissues; can include Hashimoto thyroiditis, lupus, or autoimmune thrombocytopenia, etc.

**Immunodeficiency** - Lack of an effective immune response to fight an infection or illness; can include T-cell immunodeficiency, other cellular immunodeficiencies, or combined immunodeficiency, etc.

**Increased inflammatory response** - An overreaction of the body to injury or infection that results in too much redness or swelling

Age symptoms first appeared	Age at diagnosis
Unsure	Unsure
Before birth	Before birth
0 - 3 months old	0 - 3 months old
4 - 7 months old	old
8 - 11 months old	4 - 7 months old
1 year old	old
2 years old	8 - 11

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**Recurrent infections** - Infections that "come back" more often than normal despite treatment

**Rheumatoid arthritis** - An autoimmune-caused form of arthritis; includes juvenile and adult forms

- |              |              |
|--------------|--------------|
| 3 years old  | months old   |
| 4 years old  | 1 year old   |
| 5 years old  | 2 years old  |
| 6 years old  | 3 years old  |
| 7 years old  | 4 years old  |
| 8 years old  | 5 years old  |
| 9 years old  | 6 years old  |
| 10 years old | 7 years old  |
| 11 years old | 8 years old  |
| 12 years old | 9 years old  |
| 13 years old | 10 years old |
| 14 years old | 11 years old |
| 15 years old | 12 years old |
| 16 years old | 13 years old |
| 17 years old | 14 years old |
| 18 years old | 15 years old |
| 19 years old | 16 years old |
| 20 years old | 17 years old |
| 21 years old | 18 years old |
| 22 years old | 19 years old |
| 23 years old | 20 years old |
| 24 years old | 21 years old |
| 25 years old | 22 years old |
| 26 years old | 23 years old |
| 27 years old | 24 years old |
| 28 years old | 25 years old |
| 29 years old | 26 years old |
| 30 years old | 27 years old |
| 31 years old | 28 years old |
| 32 years old | 29 years old |
| 33 years old | 30 years old |
| 34 years old | 31 years old |
| 35 years old | 32 years old |
| 36 years old | 33 years old |
| 37 years old | 34 years old |
| 38 years old | 35 years old |
| 39 years old | 36 years old |
| 40 years old | 37 years old |
| 41 years old | 38 years old |
| 42 years old | 39 years old |
| 43 years old | 40 years old |
| 44 years old | 41 years old |
| 45 years old | 42 years old |
| 46 years old | 43 years old |
| 47 years old | 44 years old |
| 48 years old | 45 years old |
| 49 years old | 46 years old |
| 50 years old | 47 years old |
| 51 years old | 48 years old |
| 52 years old | 49 years old |
| 53 years old | 50 years old |
| 54 years old | 51 years old |
| 55 years old | 52 years old |
| 56 years old | 53 years old |
| 57 years old | 54 years old |
| 58 years old | 55 years old |
| 59 years old | 56 years old |
| 60 years old | 57 years old |
| 61 years old | 58 years old |
| 62 years old | 59 years old |
| 63 years old | 60 years old |
| 64 years old | 61 years old |
| 65 years old | 62 years old |
| 66 years old | 63 years old |
| 67 years old | 64 years old |
| 68 years old | 65 years old |
| 69 years old | 66 years old |
| 70 and older | 67 years old |
|              | 68 years old |
|              | 69 years old |
|              | 70 and older |

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

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

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same issue.

## Answers

**Blood clot** - Select this choice if a blood clot has occurred in one or more of your veins or arteries (thrombosis)

**Blood clotting** - Select this choice if you have a coagulation disorder, meaning an issue with your blood clotting too fast or too slow

**Easy bleeding** - Bleeding with little or no trauma

**Easy bruising** - Bruising with little or no trauma

**Persistent bleeding** - Bleeding that continues for a long period of time after trauma; difficulties getting bleeding to stop once it has started

**Extramedullary hematopoiesis** - A specific blood issue where blood cells form and develop outside of the bone marrow

**Frequent nosebleeds** - Frequent occurrence of nosebleeds (epistaxis)

**Platelets** - An issue with too many or too few platelets (thrombocytosis or thrombocytopenia)

**Red blood cells** - An issue with red blood cells, leading to problems such as anemia, polycythemia, or thalassemia, etc.

**White blood cells** - An issue with too many or too few white blood cells (leukocytosis or leukopenia)

## Age symptoms first appeared

Unsure

Before birth

0 - 3 months old

4 - 7 months old

8 - 11 months old

1 year old

2 years old

3 years old

4 years old

5 years old

6 years old

7 years old

8 years old

9 years old

10 years old

11 years old

12 years old

13 years old

14 years old

15 years old

16 years old

17 years old

18 years old

19 years old

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36 years old

37 years old

38 years old

39 years old

40 years old

41 years old

42 years old

43 years old

44 years old

45 years old

46 years old

47 years old

48 years old

49 years old

50 years old

51 years old

52 years old

53 years old

54 years old

55 years old

56 years old

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58 years old

59 years old

60 years old

61 years old

62 years old

63 years old

64 years old

65 years old

66 years old

67 years old

68 years old

69 years old

70 and older

## Age at diagnosis

Unsure

Before birth

0 - 3 months old

old

4 - 7 months old

old

8 - 11 months old

old

1 year old

2 years old

3 years old

4 years old

5 years old

6 years old

7 years old

8 years old

9 years old

10 years old

11 years old

12 years old

13 years old

14 years old

15 years old

16 years old

17 years old

18 years old

19 years old

20 years old

21 years old

22 years old

23 years old

24 years old

25 years old

26 years old

27 years old

28 years old

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35 years old

36 years old

37 years old

38 years old

39 years old

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41 years old

42 years old

43 years old

44 years old

45 years old

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56 years old

57 years old

58 years old

59 years old

60 years old

61 years old

62 years old

63 years old

64 years old

65 years old

66 years old

67 years old

68 years old

69 years old

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70 and older

**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.**

## 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?**