

*GENE*ous

GENEous

This application is an interactive visual representation of the Mendelian Inheritance in Man, a comprehensive authorities compendium of human genes and genetic phenotypes, a catalogue of genes and genetic Disorders. This catalogue contains information on all known mendelian disorders and over 12,000 genes. It focuses on the relationship between phenotype and genotype. It's meant to be used for further research and as a tool for genomic analysis.

Within the application, there are three different ways of navigating through the human genome and exploring the various genetic disorders represented. One can utilize the simplified representation of the human body to begin their exploration through genetic disorders affecting specific locations of the

body. The locations of the genes holding these disorders will then be represented on the twenty-four chromosomes. These loci of these genes will highlight and from there it is easy to see which parts of the body are affected and where in the chromosomes these genes are located. These chromosomes can be singled out and zoomed into. The chromosome is enlarged to show all of the loci of the genes and the phenotypes that they affect. This view will show the exact locations of the genes and their names. The section for affected locations helps go into depth within specific systems in the body, which are affected by genetic disorders. Every page in the application holds ways to reach all other pages. Thus, one can seamlessly navigate through the application.

Idea

OMIM Database

Chromosome

Home | About | Statistics | Downloads/API | Help | External Links | Terms of Use | Contact Us | MIMmatch **NEW**

Select Language

Search gene map Search

Search: OMIM | Advanced Search: OMIM, Clinical Synopses, OMIM Gene Map | Search History: View, Clear | Toggle: **Phenotype Only Entries** All Entries

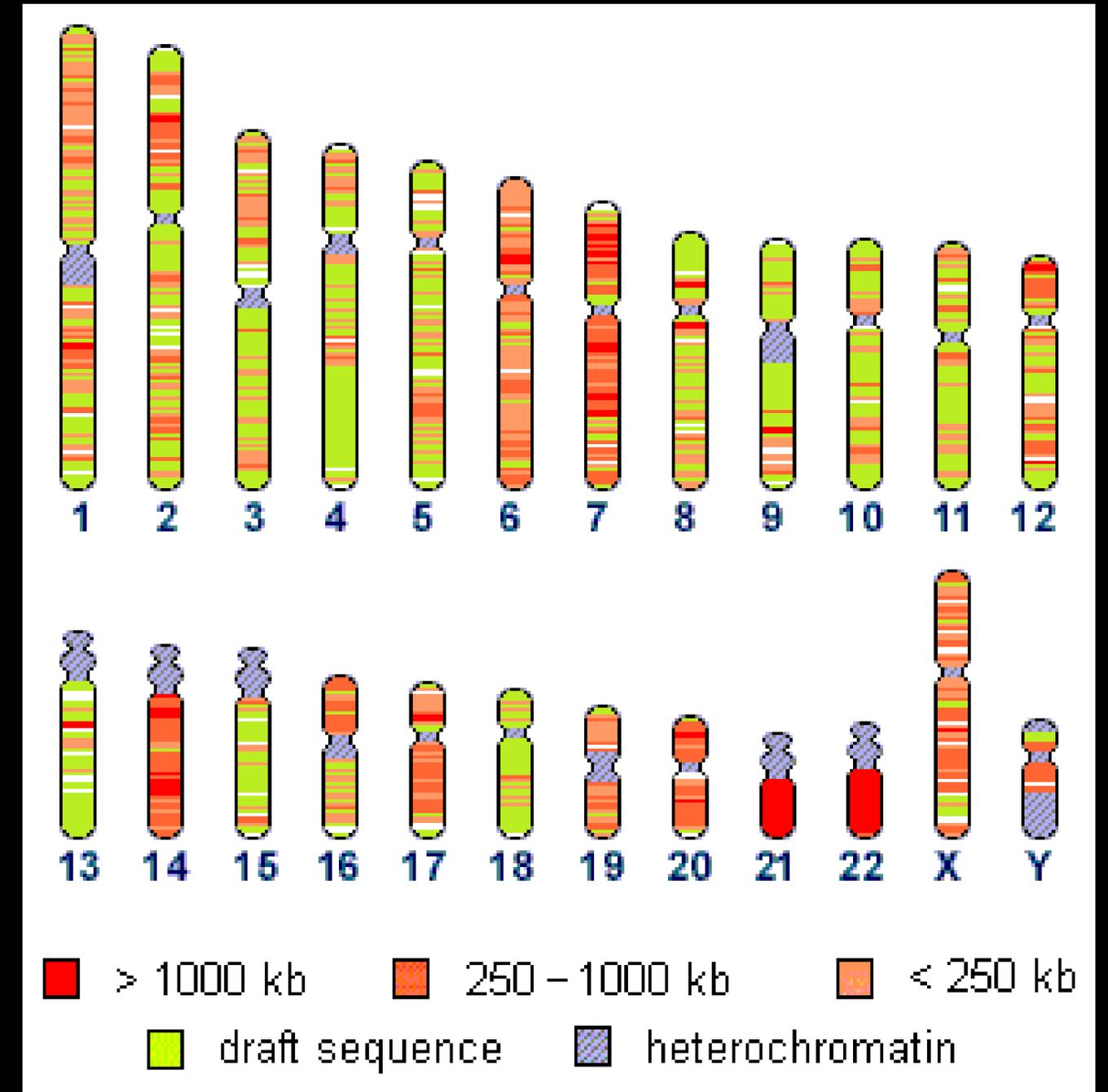
OMIM Gene/Loci: 1 - 10 of 795 on Chromosome 3 (All Entries) | Show 100

pter Towards pter Back 3 Forward 3 Towards qter qter

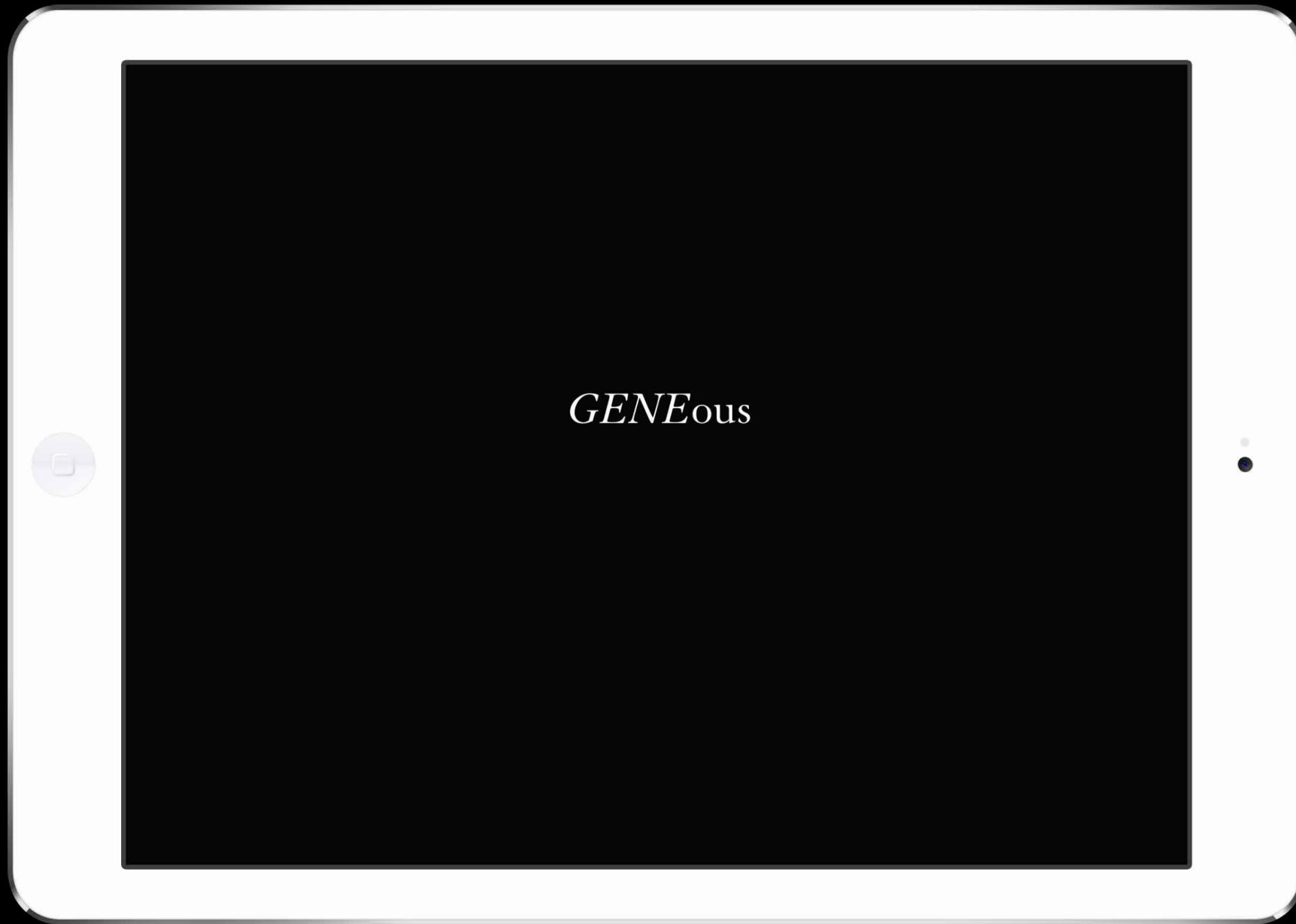
Location (genomic start, cyto location) (from NCBI)	Gene/Locus	Gene/Locus name	Gene/Locus MIM number	Phenotype	Phenotype MIM number	Pheno map key	Comments	Mouse symbol (from MGI)
3:0 3p	CRCL	Creatinine clearance QTL	607135	Creatinine clearance QTL	607135	2	in African Americans	
3:0 3pter-p25	DEL3pterp25, C3DELpterp25	3p- syndrome (chromosome 3pter-p25 deletion syndrome)	613792	3p- syndrome	613792	4	contiguous gene deletion syndrome	
3:0 3p26	HPC5	Prostate cancer, hereditary, 5	609299	{Prostate cancer, hereditary, 5}	176807	2	between D3S1270 and D3S4559	
3:0 3p26	IBD9	Inflammatory bowel disease 9	608448	{Inflammatory bowel disease 9}	608448	2		
3:0 3p26-p24.2	MYMY1, MYMY	Moyamoya disease	252350	Moyamoya disease	252350	2	max lod at D3S3050	
3:0 3p26	STQTL5	Stature quantitative trait locus 5	608982	{Stature QTL 5}	608982	2	max lod between D3S1297 and D3S1304	
3:238,278 3p26.3	CHL1, CALL, L1CAM2	CHL1, mouse, homolog of (L1 cell adhesion molecule 2)	607416					Chl1
3:1,134,341 3p26.3	CNTN6, NB3	Contactin 6	607220					Cntn6
3:2,140,549 3p26.3-p26.2	CNTN4	Contactin 4	607280					Cntn4
3:3,108,007 3p26.2	IL5RA	Interleukin-5 receptor, alpha	147851					Il5ra

pter Towards pter Back 3 Forward 3 Towards qter qter

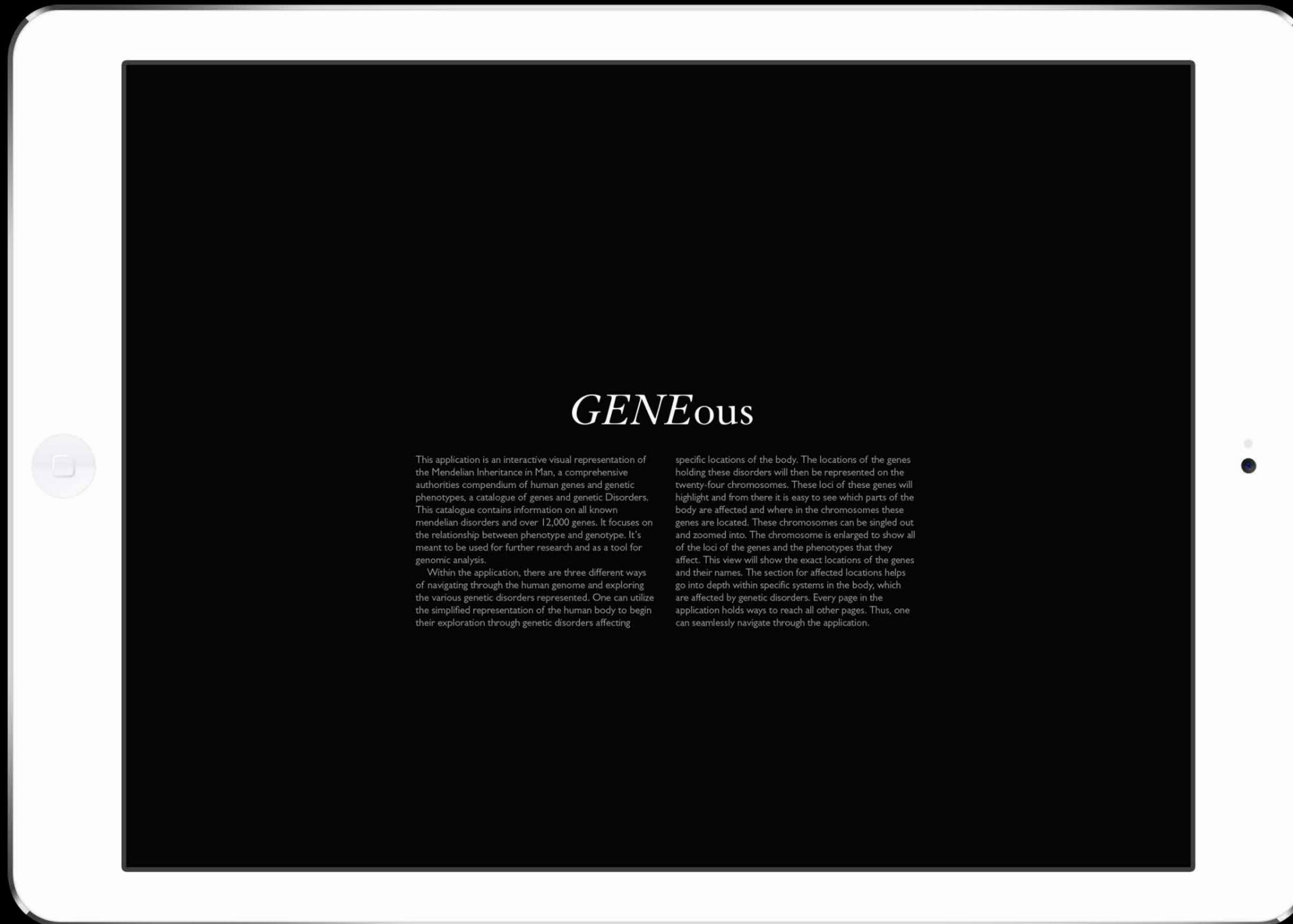
NOTE: OMIM is intended for use primarily by physicians and other professionals concerned with genetic disorders, by genetics researchers, and by advanced students in science and medicine. While the OMIM database is open to the public, users seeking information about a personal medical or genetic condition are urged to consult with a qualified physician for diagnosis and for answers to personal questions.



User Story

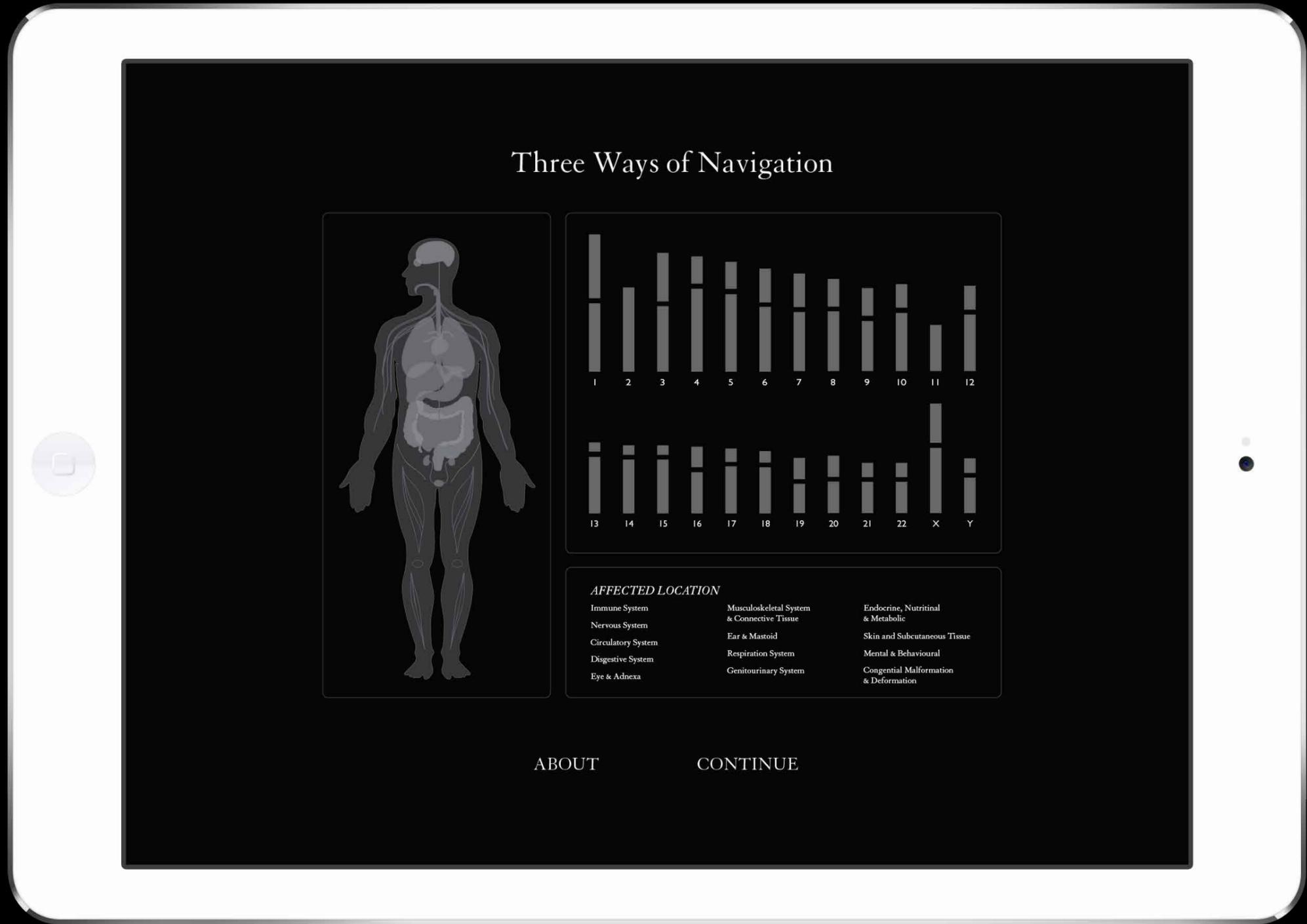


1 User opens the app.



2 Application introduction automatically appears. User taps to continue.

About page for first time users only. The about page provides background information on the app and how the navigation works.



3 User taps the image area to read descriptions on the three ways of navigation.

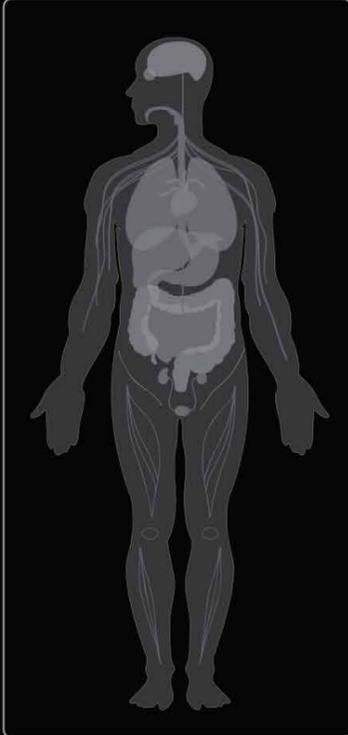
Navigation description page for first time users only. This page shows the three ways users are able to navigate through the app.

The Body, Chromosomes, and Locations sections can be selected to bring up more information on each of the navigation system.

This page gives users the option to view the app description or to enter the app.

4 User taps to continue to the next navigation tool.

By the Body



The human body is made up of millions of cells each specializing in a particular function. Humans have two types of chromosomes—sex chromosomes and autosomes.

AFFECTED LOCATION

Immune System	Musculoskeletal System & Connective Tissue	Endocrine, Nutritional & Metabolic
Nervous System	Ear & Mastoid	Skin and Subcutaneous Tissue
Circulatory System	Respiratory System	Mental & Behavioural
Digestive System	Genitourinary System	Congenital Malformation & Deformation
Eye & Adnexa		

ABOUT CONTINUE

5

User taps to continue to the next navigation tool.

By the Chromosomes

A chromosome is a structure of DNA, protein and RNA found in cells. It is a single piece of coiled DNA containing many genes. A gene is the molecular unit of heredity of a living organism. Genes hold the information to build and maintain an organism's cells and pass genetic traits to offspring.



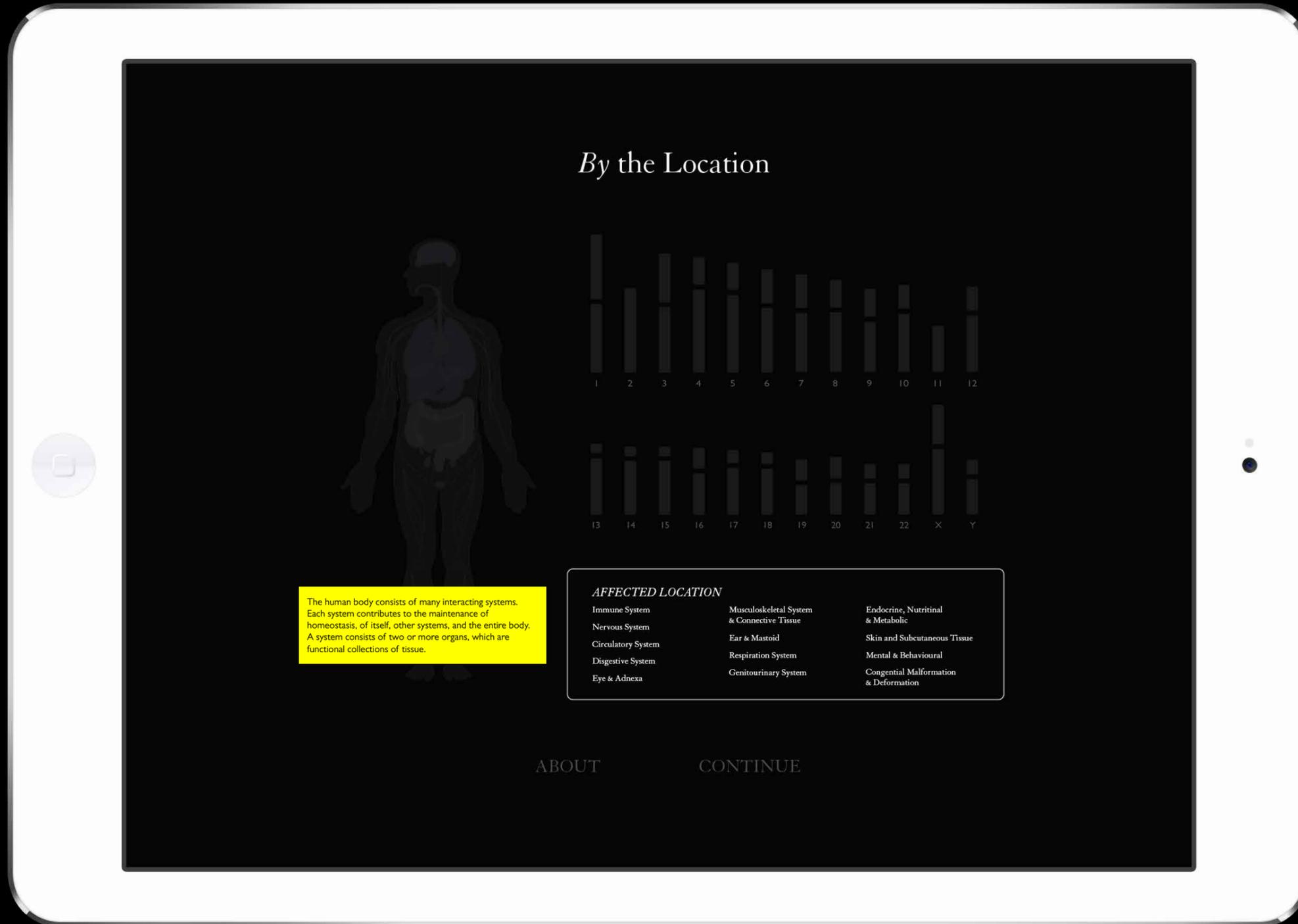
AFFECTED LOCATION

Immune System	Musculoskeletal System & Connective Tissue	Endocrine, Nutritional & Metabolic
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Eye & Adnexa		

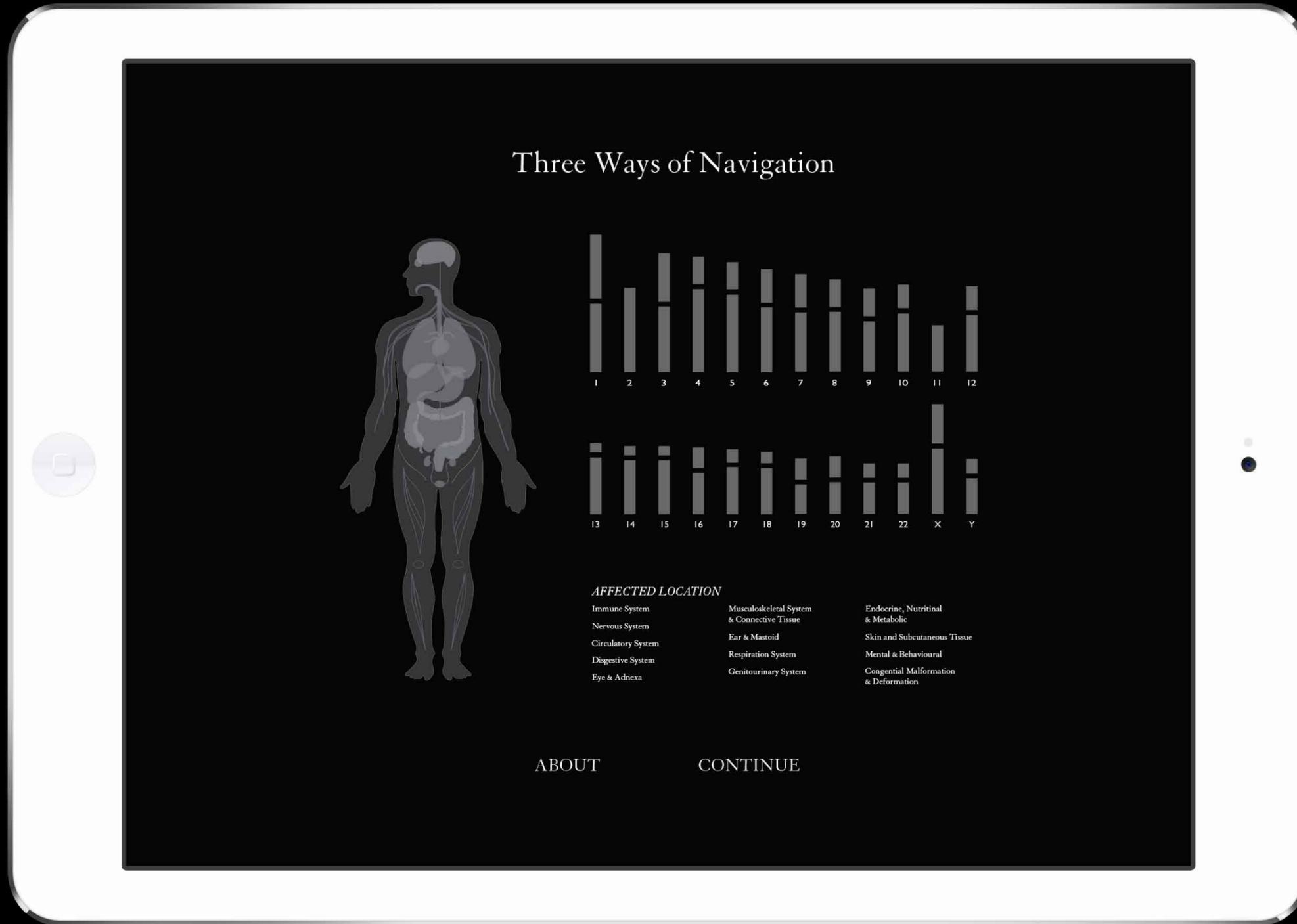
ABOUT

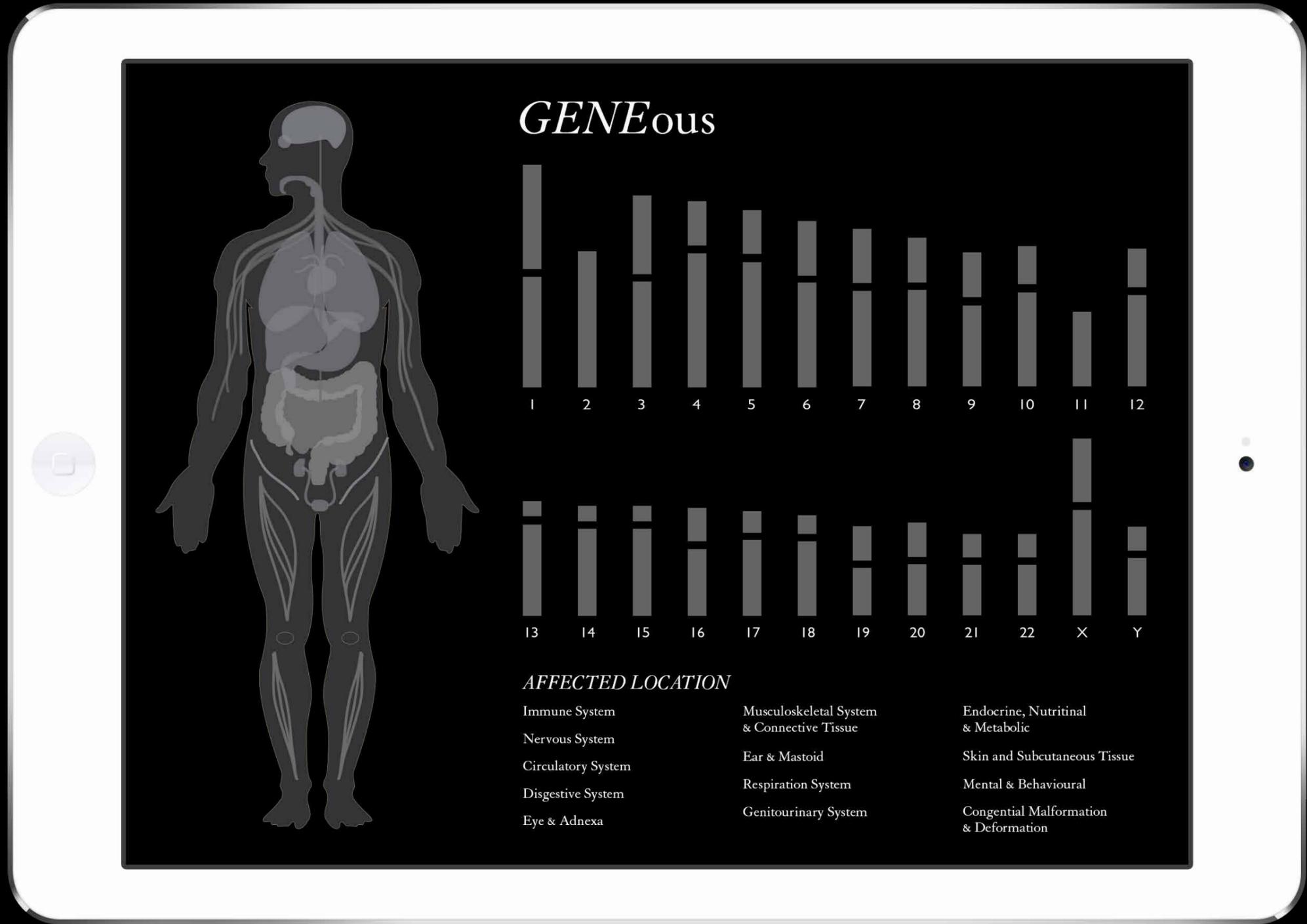
CONTINUE

6 User taps to continue to exit the explanations.



7 User taps to **Continue** to enter the application.

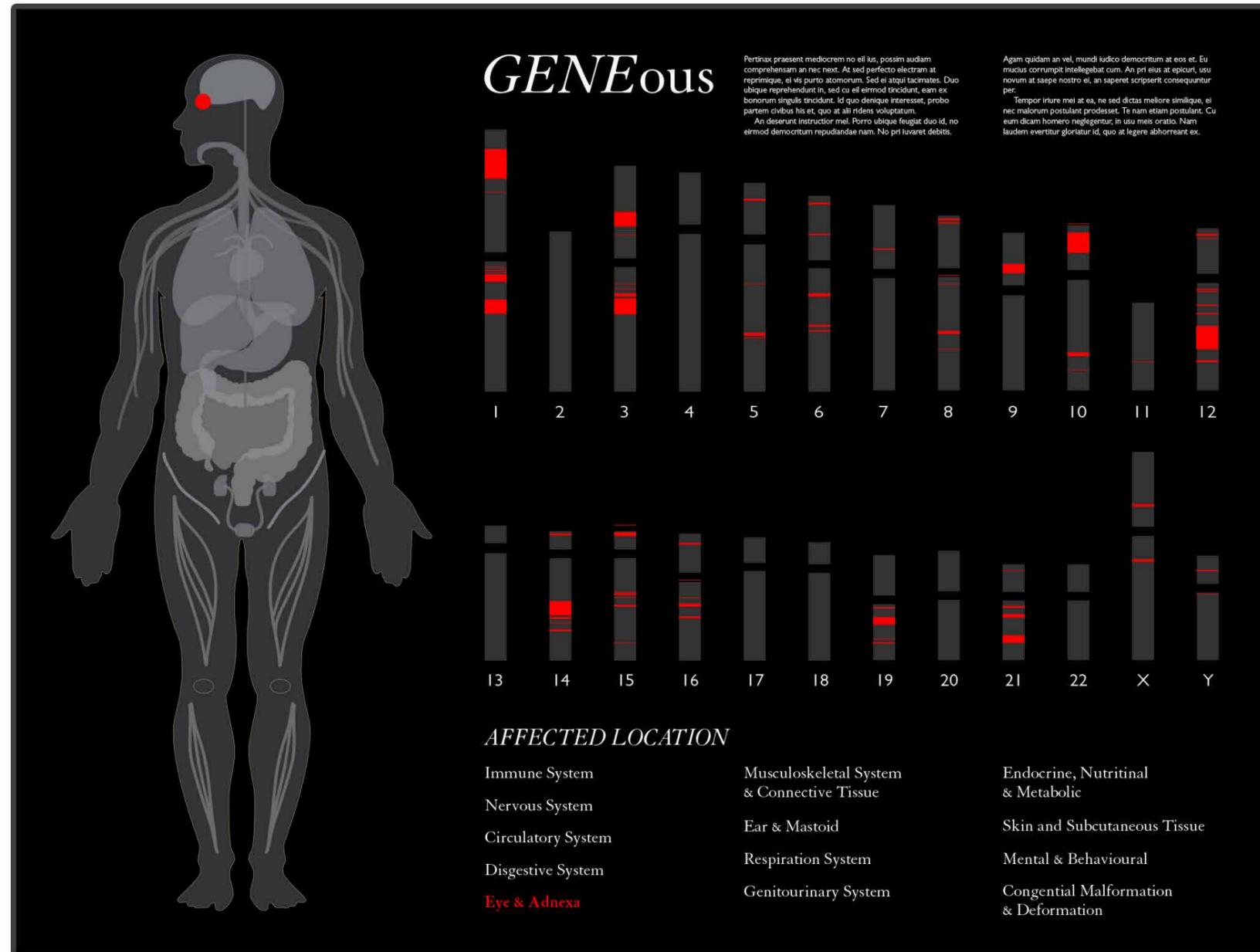




8 User selects **Eye & Andea** under **Affected Locations**.

This page features the previously shown three ways of navigation. From here users can begin to use the app in the way that is most efficient them.

The body focuses on the physical locations the disorders affect. The affected locations focuses on the body systems that are affected by the genetic disorder.



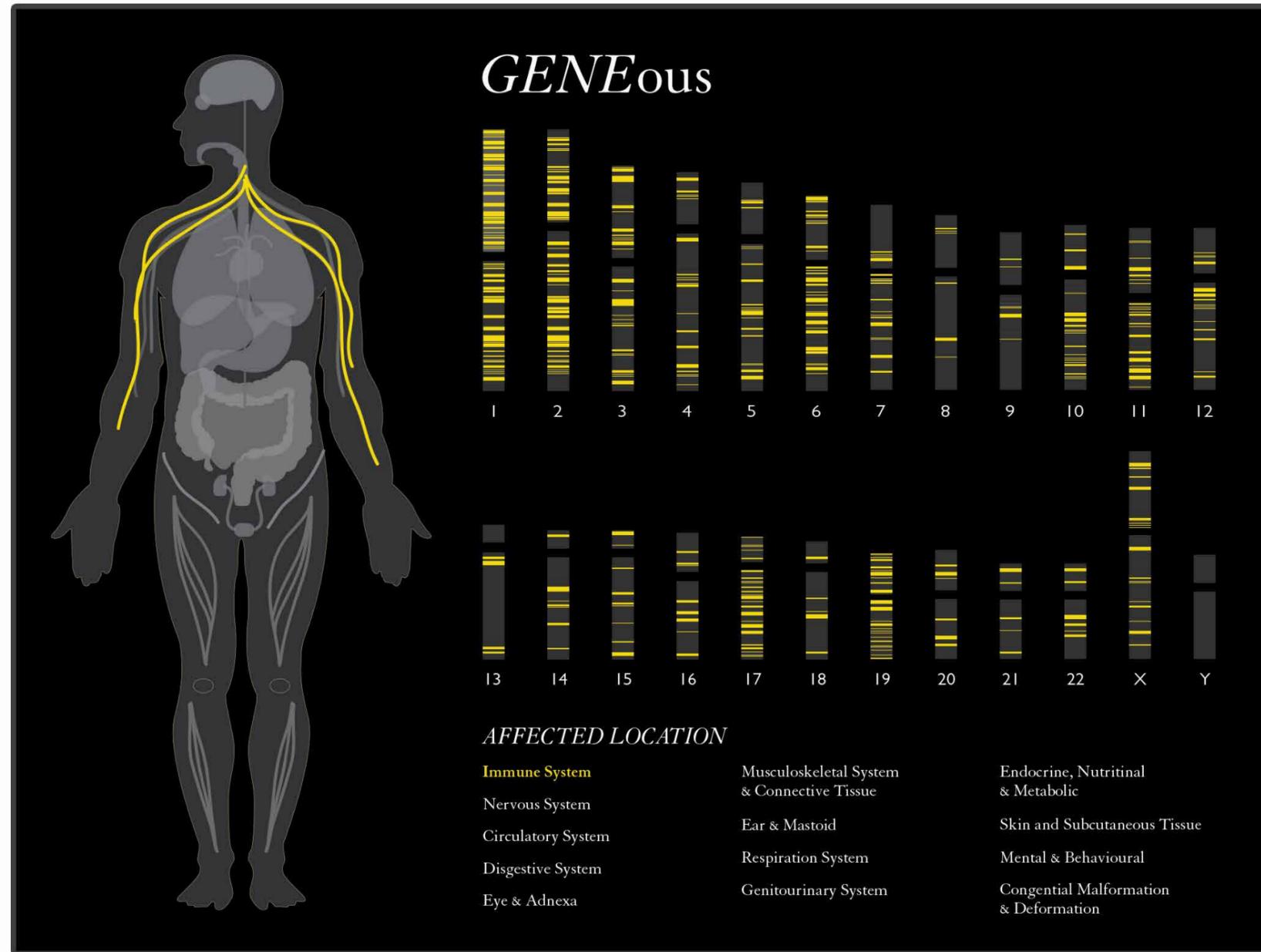
9 User selects **Immune System** under **Affected Locations**.

This pages shows how the app page would look once the user has selected the “Eye & Adnea” from the Affected Locations list.

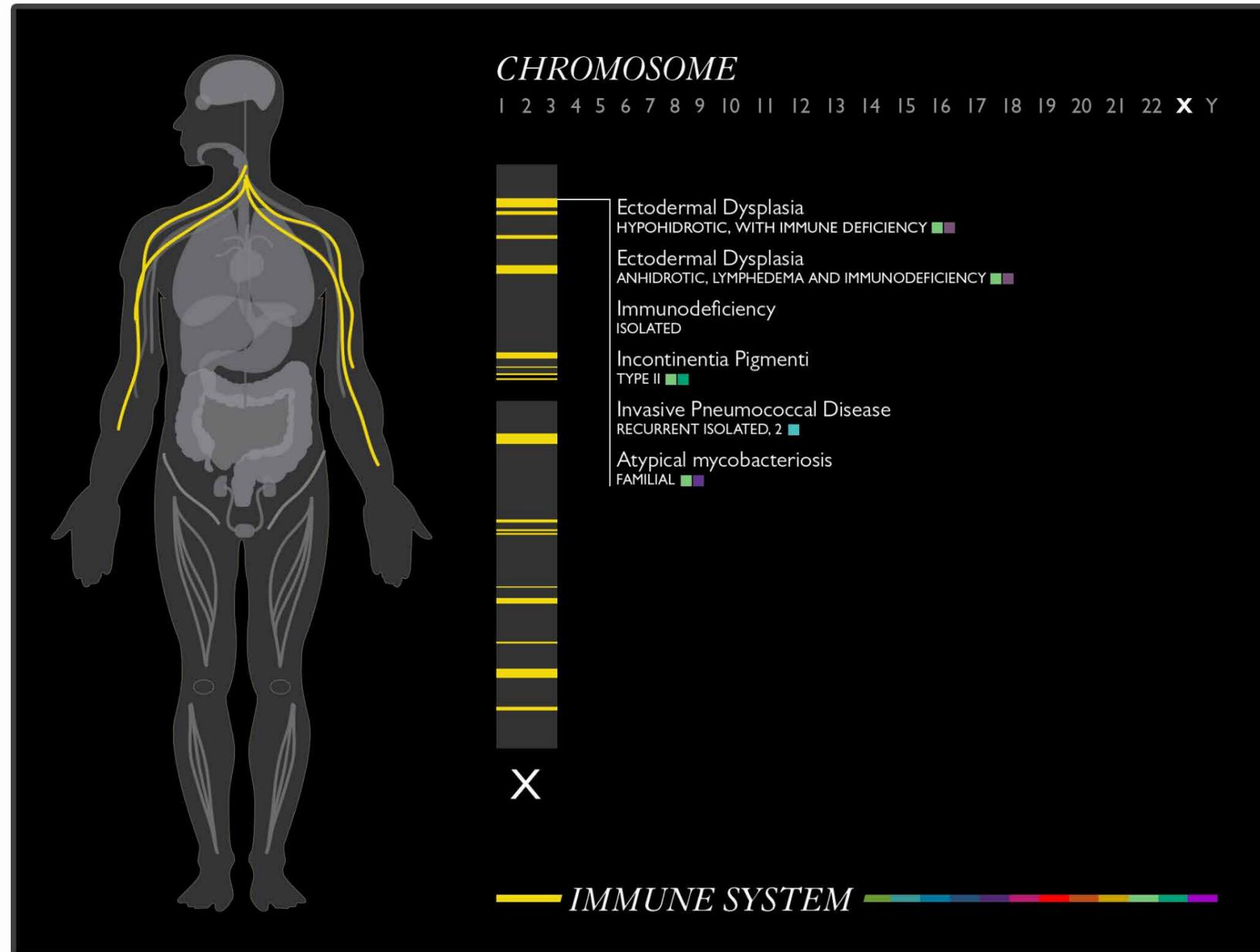
The “Eye & Adnea” is highlighted to show what the user is looking at. The eye is highlighted on the body to show where in the actual human body the disorders affect.

Every gene that affects the eye and adnea is highlighted on the chromosomes.

10 User selects the **X** chromosome under the **Chromosome** section.



11 User selects the **Ectodermal Dysplasia**, a specific genetic disorder.



This page shows the view of the specific chromosome chosen and the location of the gene on the chromosome.

From this page the use can select a specific gene on the chromosome to see the specific disorder that it affects. The color blocks next the disorders show the other locations the disorder affects.

From this page the user can navigate to other chromosomes or to another affected locations or body systems.

Ectodermal Dysplasia
HYPOHIDROTIC, WITH IMMUNE DEFICIENCY

AFFECTED LOCATION

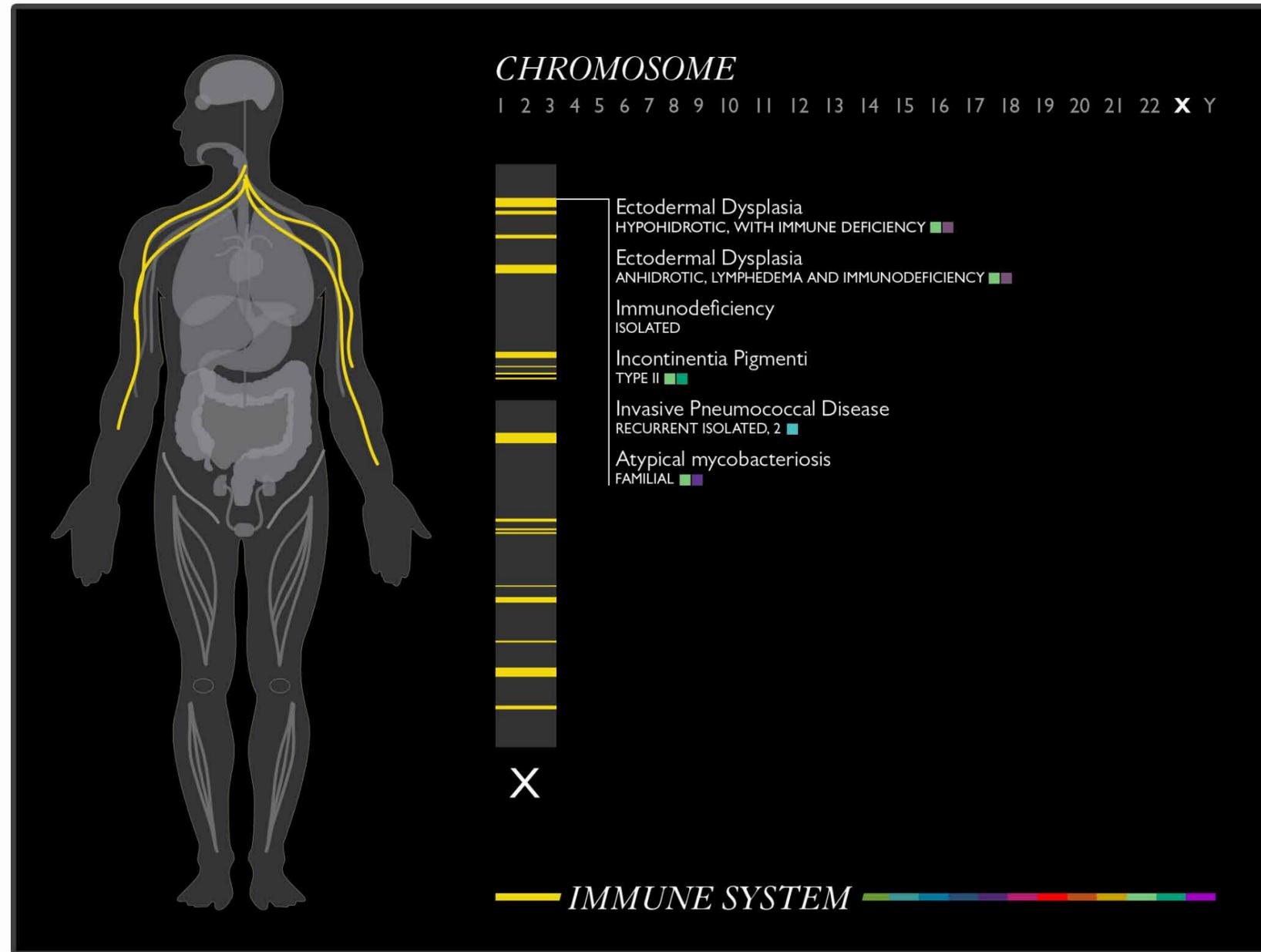
Immune System	Musculoskeletal System & Connective Tissue	Endocrine, Nutritional & Metabolic
Nervous System	Ear & Mastoid	Skin and Subcutaneous Tissue
Circulatory System	Respiration System	Mental & Behavioural
Digestive System	Genitourinary System	Congenital Malformation & Deformation
Eye & Adnexa		

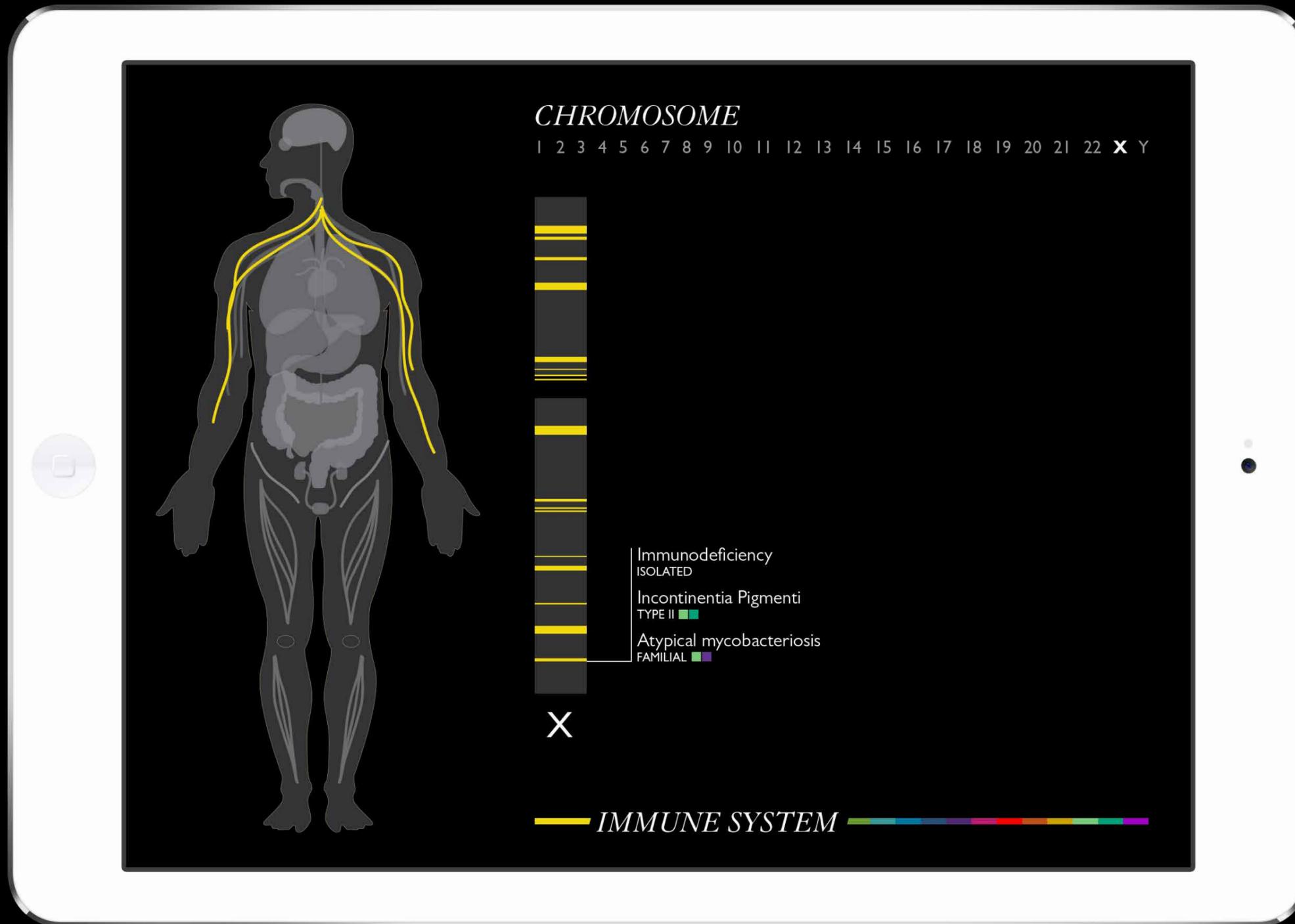
12 User selects the **X chromosome**.

The page shows the main navigation page.

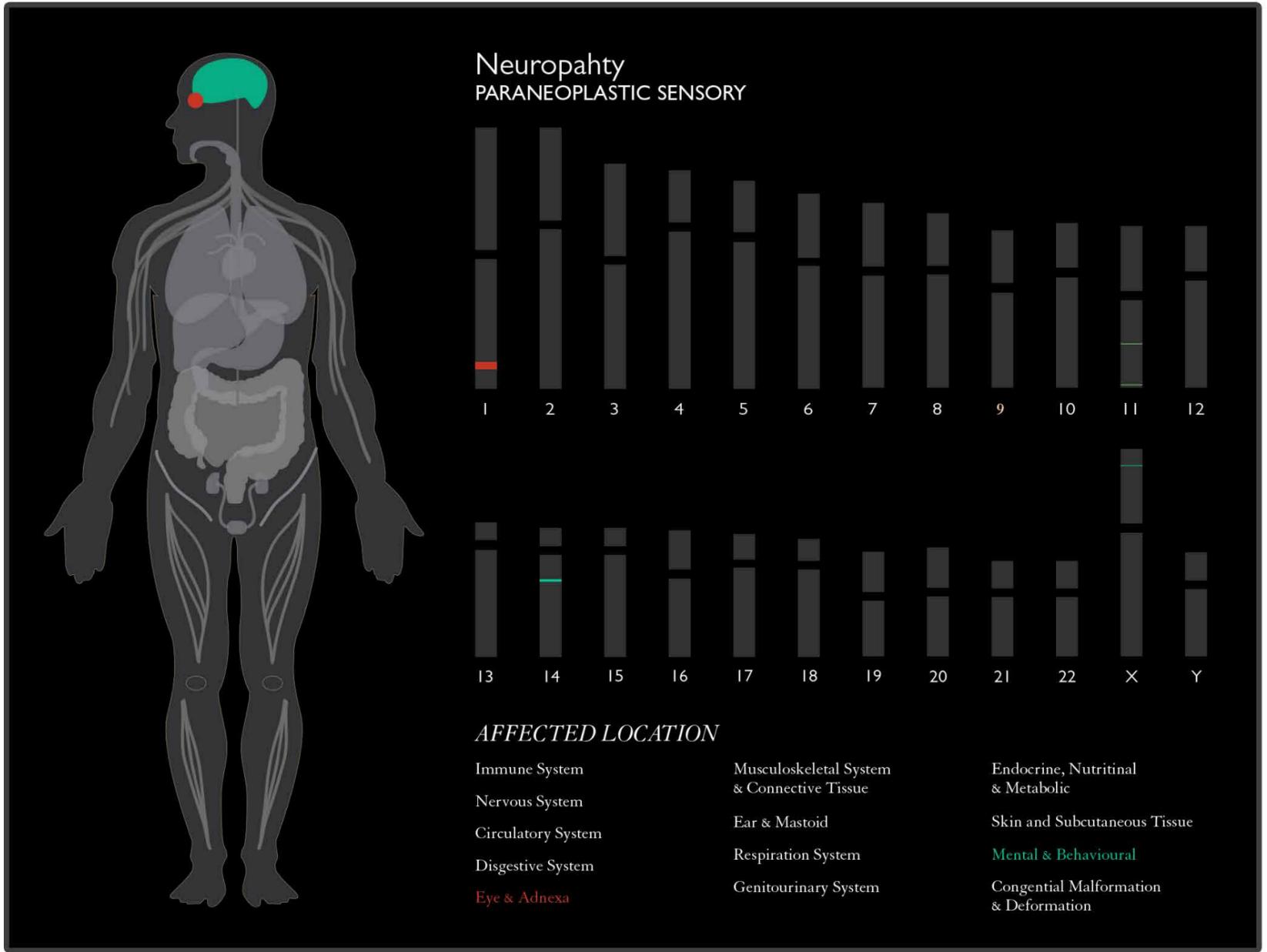
The systems of the body that are affected are highlighted on the body. The genes affecting/ causing Ectodermal Dysplasia are highlighted in the chromosomes. The affected locations of Ectodermal Dysplasia are highlighted.

13 User selects the **last highlighted bar** on the **X chromosome**.





14 User selects the **first highlighted bar** on the **X chromosome**.



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Topic Information Design
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