CLIENT NAME
Day Month 2016

Research Highlights

GOALS

- Identify the segments of DNA that Paul XXXX inherited from each of his grandparents and trace the inheritance of specific traits.

PROGRESS

- Mapped 96.7% of Paul's genome to each of his four grandparents.
- Discovered that Paul shares the following percentages of DNA with his grandparents: 27.4% with Lulu, 20.2% with Alan, 23.8% with Richard and 25.3% with Arlene (3.3% was not mapped).
- Identified the inheritance patterns for genes affecting physical and behavioral characteristics, including eye, hair and skin color; nose, eye, ear, mouth, hand, teeth and face morphology; hair, eyebrow and beard shape; balding patterns; musical ability; smell, taste and color vision.

RECOMMENDATIONS

1. Continue to map Paul's chromosome through collaboration with genetic cousins, direct mapping, and indirect mapping to identify the deeper origin of specific traits and to map segments to previous generations of Paul's ancestry.
2. Use genetic genealogy and traditional genealogy to discover more about Paul's ancestors.

Research Report

DNA and Inheritance

Before exploring the details of Paul's DNA and genetic inheritance, it is useful to understand the general nature of DNA and inheritance. DNA determines many physical, behavioral, emotional, and mental characteristics, as well as how the human body performs certain functions and processes. It is responsible for the inheritance of genetic traits and is the foundational code which is used to create, maintain and regulate all types of cells and all types of life. Strands of molecules form the double helix structure of DNA, and these long strands of DNA are organized into compact structures called chromosomes. Each human has 23 pairs of chromosomes – 22 autosomal chromosomes and one pair of sex chromosomes – which together are composed of more than 3.2 billion base-pairs of DNA.

Each individual inherits one set of chromosomes from their mother (50%) and one set of chromosomes from their father (50%). These chromosome pairs are not exactly identical, but are homologous, meaning that for every chromosome inherited from an individual's mother, they have inherited another chromosome of the same size, shape, and organization of genetic material from their father. Occasionally mutations occur when DNA is replicated resulting in slight differences between two copies of homologous chromosomes.

DNA can be divided into functional units called genes, and each person has two copies of each gene which may differ slightly from one another. These different copies are known as alleles. Though it may be tempting to say that an individual got Grandpa's nose or Grandma's hair, each individual inherits two alleles from at least two different grandparents, though one allele may be more dominant than the other. Many traits are determined by multiple genes, making identification of the origins of a specific trait quite difficult. On the other hand, some genes produce proteins which affect multiple traits.

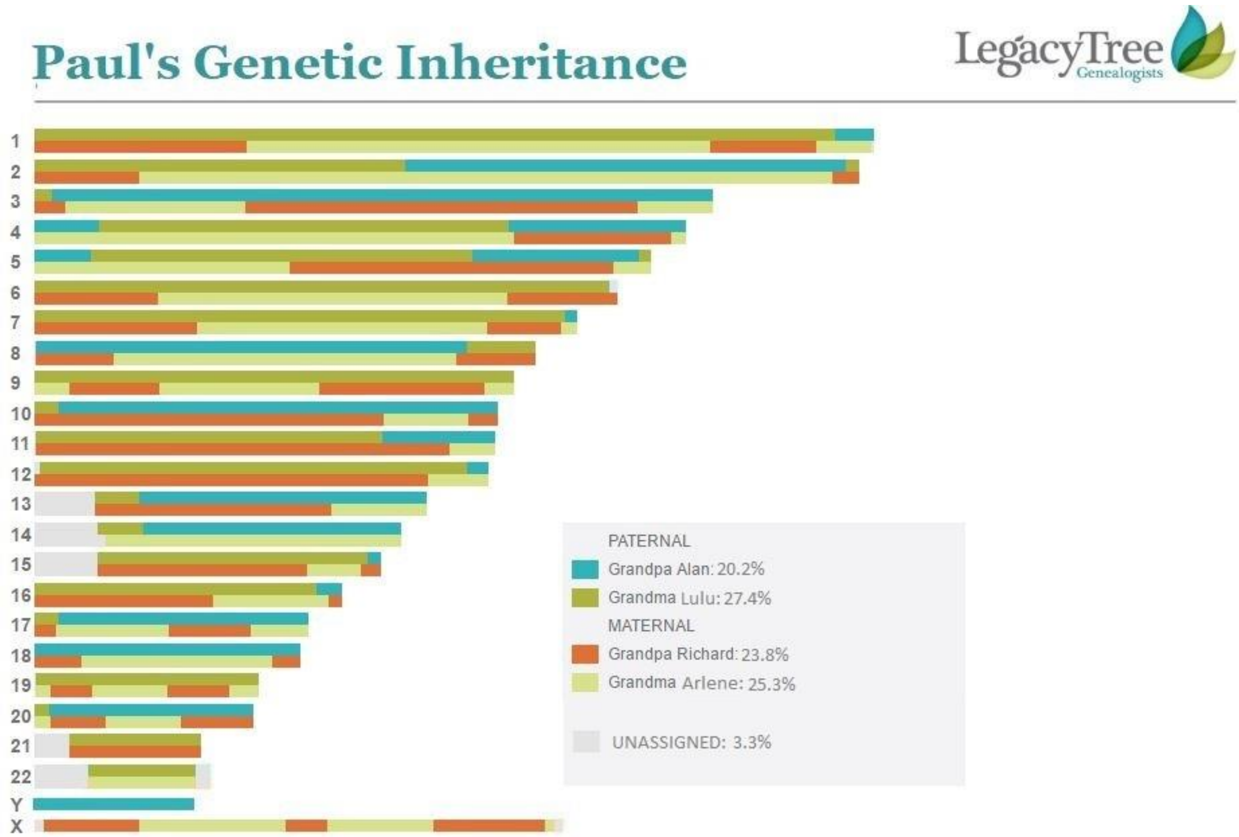
Though some genes that affect physical traits are known, there are many other genes which may affect the expression of certain traits that are as of yet undiscovered.

Paul's Genetic Inheritance

In this project, we were asked to identify the segments of DNA that Paul inherited from each of his grandparents. We were able to do this by performing genetic testing on Paul, his paternal grandmother, and his maternal grandmother, and then using proprietary software to infer inheritance from the remaining grandparent on each side.

Through this process we identified each DNA segment that Paul inherited from each of his grandparents. Since each individual has a maternally and a paternally inherited chromosome, any section of maternal DNA where Paul does not match his maternal grandmother has to have come from his maternal grandfather. Likewise, any section of paternally inherited DNA where Paul did not match his paternal grandmother had to have come from his paternal grandfather.

The following genetic inheritance chart shows the segments of DNA that Paul inherited from each of his grandparents. From this we see that on the paternal side Paul inherited the most DNA from his grandmother, Lulu, and on the maternal side, he inherited the most DNA from his grandmother, Arlene. Some segments of DNA are not tested in genetic genealogy tests due to a lack of unique markers or unreliable results in those areas. Also some segments of DNA were too small to confidently assign to a specific ancestor. These segments are included in the unassigned categories and amount to 4% of Paul's genome.



Having identified the segments of DNA that Paul inherited from his grandparents, we were able to identify the genes associated with these segments and their resulting traits. It should be kept in mind that some traits are determined by multiple genes, while others are determined by a single gene. On the other hand, some genes affect multiple traits whereas other genes only have a single function. Also, in this report we have only included genes which have a known effect in normal variation of traits. We have not included genes whose mutations result in differences due to rare disorders.

Following are several tables which report genes associated with specific physical characteristics. For those genes where multiple associations have been made to SNPs (DNA variations) within their coding regions, we have reported the number of SNPs associated with those traits and the range of the entire gene. For those genes where only one SNP has been found to be associated, we have reported the site of the SNP and the possible gene implicated. Wherever possible, we have also provided a description of the effect of variation in that gene.

For traits which are governed by multiple genes, we have highlighted in **green** those that have the strongest association and the strongest effect. Finally, if the majority of the genes for a particular trait came from one of Paul's grandparents, we have highlighted in **teal** which individual contributed the largest number of genes relating to that specific trait.

Chart key

Gene or location: region of the genome where variant(s) are found.
SNP: A single nucleotide polymorphism. A common mutation at a single site from one base to another.
Chr: Chromosome number
Start: The starting point of the gene named. In the case of a mutation, the start and stop point will be the same.
Stop: The end point of the gene or mutation named. In the case of a mutation, the start and stop point will be the same.
PGP: Paternal Grandparent.
MGP: Maternal Grandparent.
Description: Effect of the gene variant on a trait.

Eye Color

To date, scientific research has identified more than 30 sites in the genome associated with eye color variation. Two genes, HERC2 and OCA2, account for the majority of this variation while four more genes account for significant variation. Several other genes play smaller roles in color determination.[1] In this case, Paul inherited the majority of his known eye color genes from Lulu and from Richard and he also inherited the most significant determinants of eye color from these same two individuals.

Gene or location	SNPs (# / name)	Chr	Start	Stop	PGP	MGP	Description
HERC2	18	15	28356183	28567313	Lulu	Richard	brown vs. blue
OCA2	12	15	28000021	28344483	Lulu	Richard	blue, brown, green, hazel
SLC24A4	4	14	92788925	92967825	Alan	Arlene	
SLC45A2	2	5	33944721	33984780	Lulu	Arlene	lighter vs. darker
TYR	7	11	88911040	89028927	Lulu	Richard	
IRF4	2	6	391739	411443	Lulu	Richard	
TYRP1	10	9	12693385	12710285	Lulu	Richard	
LYST	2	1	235824331	236047008	Lulu	Arlene	
NPLOC4	2	17	79523913	79604138	Alan	Arlene	
TTC3	4	21	38445554	38575408	Lulu	Richard	
DSCR9	rs7277820	21	38580309	38580309	Lulu	Richard	

Eye Shape

Eye shape is determined by the configuration of skin, fat, bone, connective tissue and muscle around the eyeball. Though there are many mutations which are known to have an effect on eye appearance, many of these are likely secondary and little research has been performed to determine which genes directly contribute to eye shape. It is likely that there

[1] Fan Liu, et al., "Eye color and the prediction of complex phenotypes from genotypes," in *Current Biology,* Vol 19:5 (10 March 2009), R192-R193, www.sciencedirect.com, accessed July 2016; and,

Fan Liu, et al., "Digital Quantification of Human Eye Color Highlights Genetic Association of Three New Loci," in *PLOS Genetics,* 6 May 2010, journals.plos.org, accessed July 2016.

are many genes which contribute in small ways to eye shape. We found only one study that drew connections between specific genes and eye shape based on the amount of sagging in the eyelid.[2]

Gene or location	SNPs (# / name)	Chr	Start	Stop	PGP	MGP	Description
DLGAP1	3	18	3496030	4455310	Alan	Richard	Associated with protection against sagging eyelids

Eyebrows

A recent study on the genetics of facial and scalp hair identified some of the genes responsible for variation in eyebrow thickness and the presence of monobrows.[3]

Gene or location	SNPs (# / name)	Chr	Start	Stop	PGP	MGP	Description
FOXL2	5	3	138663066	138665982	Alan	Richard	Thickness
PAX3	rs2218065	2	223034082	223034082	Alan	Arlene	Monobrow

Color Vision

Color vision in humans is dependent on three types of cones present in the retina of the eye which can detect large, medium and small wavelengths of light. Scientific research has

[2] Leoinie C Jacobs, et al., "Intrinsic and Extrinsic Risk Factors for Sagging Eyelids," in *JAMA Demortology,* Vol 150:8 (August 2014), 836-843, archderm.jamanetwork.com, accessed July 2016.

[3] Kaustubh Adhikari, et al., "A genome-wide association scan in admixed Latin Americans identifies loci influencing facial and scalp hair features," in *Nature Communications,* vol. 7 (1 March 2016) www.nature.com, accessed July 2016.

shown that mutations in the genes which encode for these cones can result in different types of color blindness. Another gene has been shown to be connected to night vision.[4]

Gene or location	**Chr**	**Start**	**Stop**	**Paternal Grandparent**	**Maternal Grandparent**	**Description**
OPN1LW	X	153409725	153409725	NA	Richard	Red
OPN1MW	X	153448085	153462352	NA	Richard	Green
OPN1SW	7	128412543	128415844	Lulu	Arlene	Blue
RHO	3	129247482	129254187	Alan	Richard	Night vision

Ear shape

Ear shape is determined by the structure of underlying cartilage. By performing genomic scans on a large group of individuals, it was discovered that the following genes and sites have an effect on variation in different elements of ear morphology.[5]

Gene or location	**SNPs**	**Chr**	**Start**	**Stop**	**PGP**	**MGP**	**Description**
LOC105378934 (CART1 Binding site)	3	1	119864997	119907180	Lulu	Arlene	Antitragus size; Folding of antihelix
EDAR	15	2	109510927	109605828	Lulu	Arlene	Ear protrusion; Helix rolling; Lobe attachment; Lobe size

---

[4] Jay Neitzz and Maureen Neitz, "The genetics of normal and defective color vision," in *Vision Research,* vol 51:7 (13 April 2011), 633-651, www.sciencedirect.com, accessed July 2016.

[5] Kaustubh Adhikari, et al., "A genome-wide association study identifies multiple loci for variation in human ear morphology," in *Nature Communications,* vol. 6 (25 June 2015) www.nature.com, accessed July 2016.

SP5	4	2	171571857	171574498	Alan	Arlene	Lobe attachment; Lobe size
MPRS22	3	3	139343993	139357128	Alan	Richard	Lobe size
LRBA	rs1960918	4	151222266	151222266	Alan	Richard	Helix rolling
LOC153910	7	6	142847592	142959026	Lulu	Richard	Lobe size
rs1619249	rs1619249	18	49190644	49190644	Alan	Arlene	Folding of antihelix

Musical Ability

Traits relating to musical ability such as perfect pitch, relative pitch and musical creativity have been shown to have a genetic factor, though many of the genes that influence these traits are yet to be discovered. Genetic inheritance of musical ability is an example of a trait that has genetic underpinnings but also has a strong environmental element. This is the case for many genetic traits. Though certain genes and mutations may lead to predisposition or increased likelihood of specific traits, much of the expression of those traits also depends on environmental factors from conception to adulthood.[6]

Gene or location	**SNPs**	**Chr**	**Start**	**Stop**	**PGP**	**MGP**	**Description**
ASAP1	rs3057	8	13129837	13129837	Alan	Richard	Absolute pitch in Europeans
6q14.1-6q16.1		6	79577293	100054910	Lulu	Arlene	Absolute pitch
2q	rs1482308	2	155929442	155929442	Alan	Arlene	Absolute pitch and synaesthesia
4q22	D4S2460-D4S423	4	89833662	92473038	Lulu	Arlene	Pitch and rhythm perception
AVPR1A		12	63536539	63547971	Lulu	Richard	Pitch and rhythm

[6] Yi Ting Tan, et al., "The genetic basis of music ability," in *Frontiers in Psychology,* vol 5 (2014), 658, www.ncbi.nlm.nih.gov, accessed July 2016.

							perception; Music Memory; Music listening
rs9854612	rs9854612	3	128257214	128257214	Alan	Richard	Pitch and rhythm perception
rs13146789	rs13146789	4	30668535	30668535	Lulu	Arlene	Pitch and rhythm perception
rs13109270	rs13109270	4	30640272	30640272	Lulu	Arlene	pitch and rhythm perception
rs12510781	rs12510781	4	115640581	115640581	Lulu	Arlene	Pitch accuracy/ singing
4q23	D4S2986	4	99657666	99657892	Lulu	Arlene	Singing and pitch accuracy
UGT8	rs4148254	4	115544713	115544713	Lulu	Arlene	Singing and pitch accuracy
SLC6A4	5-HTTLPR	17	28521337	28562986	Alan	Arlene	Choir participation, Music memory

Nose Shape

The shape of the human nose is largely dependent on the structure of cartilage, bone, and muscle throughout the face. Several recent studies have found genome-wide associations to some genes that affect normal variation in nose morphology.[7] In this case Paul inherited an equal number of nose shape genes from each of his grandparents.

[7] Kaustubh Adhikari, et al., "A genome-wide association scan implicates DCHS2, RUNX2, GLI3, PAX1 and EDAR in human facial variation," in *Nature Communications,* vol. 7 (19 May 2016) www.nature.com, accessed July 2016; and,

Fan Liu, et al., "A Genome-Wide Association Study Identifies Five Loci Influencing Facial Morphology in Europeans," in *PLOS Genetics* (13 September 2012), journals.plos.org, accessed July 2016.

Gene or location	SNPs	Chr	Start	Stop	PGP	MGP	Description
DCHS2	6	4	155155527	155412930	Alan	Richard	Columelle inclination; Nose protrusion; Nose tip angle
SUPT3H/ RUNX2	5	6	44794467	45518819	Lulu	Arlene	Nose bridge breadth
GLI3	6	7	42000547	42277469	Lulu	Richard	Nose wing breadth
PAX1	4	20	21686297	21699124	Alan	Arlene	Nose wing breadth
PAX3	3	2	223064606	223163715	Alan	Arlene	Nasion position
PRDM16	5	1	2985565	3355185	Lulu	Richard	Distance from right and left alares to the pronasale

Smell

Our sense of smell is possible due to a large number of olfactory receptors which detect microscopic particles and send signals to our brain. These receptors are proteins encoded by our genes. However, the number of receptors vastly under represents the wide range of possible smells. Therefore, most receptors probably interact with a large number of molecules and whenever there may be a defective receptor, other receptors may make up for the difference. Nevertheless, there are a few genes which have been shown to have significant effects on our ability to smell specific compounds or our perception of different smells.[8]

Gene or location	**SNPs**	**Chr**	**Start**	**Stop**	**PGP**	**MGP**	**Description**
OR2J3	Multiple	6	29079587	29080661	Lulu	Richard	Cut grass
OR7D4	2	19	9324526	9325547	Lulu	Richard	Perception of androstenone
OR5A1	rs6591536	11	59211188	59211188	Lulu	Richard	β-ionone
OR2M7	rs4481887	1	248496863	248496863	Alan	Arlene	Asparagus metabolites in urine
OR11H7		14	20697562	20698503	Alan	Richard	isovaleric acid (sweat odor)
AFM295YE5		4	163600000	169200000	Alan	Richard	Pleasantness of cinnamon
OR10A2/ OR2AG2/ OR6A2	rs72921001	11	6889648	6889648	Lulu	Richard	Taste/ smell of cilantro

[8] Danielle Renee Reed and Antti Knaapila, "Genetics of Taste and Smell: Poisons and Pleasures," in *Progress in Molecular Biology and Translational Science,* Vol 94 (2010) 213-240, www.ncbi.nlm.nih.gov, accessed July 2016; and,

Nicholas Eriksson, et al., "A genetic variant near olfactory receptor genes influences cilantro preference," arXiv.org, accessed July 2016.

Mouth/ Lip Shape

While investigating the genetics of facial morphology, investigators discovered one gene that has a significant effect on variation in lip shape.[9]

Gene or location	SNPs	Chr	Start	Stop	PGP	MGP	Description
IRF6	rs642961	1	209989270	209989270	Lulu	Richard	lip protrusion and thickness

Taste

As with smell, our sense of taste is governed by receptor proteins which can detect microscopic molecules and generate a neural response. Smell and taste are also connected and influence each other. Taste receptors are frequently able to interact with many different compounds to result in different responses, and if there are defective versions of receptors, others might take over their role. Nevertheless, there are some specific genes that have been shown to have an effect on taste and taste perception.[10]

Gene or location	SNPs	Chr	Start	Stop	PGP	MGP	Description
TAS2R38	3	7	141672431	141673573	Lulu	Richard	PTC phenyl-thiocarbamide
rs307355	rs307355	1	1265154	1265154	Lulu	Richard	Sucrose sensitivity variation

[9] Shouneng Peng, et al., "Detecting Genetic Association of Common Human Facial Morphological Variation Using High Density 3D Image Registration," in *PLOS Computational Biology* (5 December 2013), journals.plos.org, accessed July 2016.

[10] Danielle Renee Reed and Antti Knaapila, "Genetics of Taste and Smell: Poisons and Pleasures," in *Progress in Molecular Biology and Translational Science,* Vol 94 (2010) 213-240, www.ncbi.nlm.nih.gov, accessed July 2016.

rs35744813	rs35744813	1	1265460	1265460	Lulu	Richard	Sucrose sensitivity variation
TAS1R3	3	1	1266726	1269844	Lulu	Richard	Umami perception
TAS1R1	3	1	6614987	6639817	Lulu	Richard	Umami perception
MGLUR1	4	6	146758782	146758782	Lulu	Richard	Umami perception
OR10A2/ OR2AG2/ OR6A2	rs72921001	11	6889648	6889648	Lulu	Richard	Taste/ smell of cilantro

Teeth

Many of the same genes that lead to the development of teeth also lead to the development of other structures like hair. Researchers investigated genes that were previously associated with hair shape and discovered that several of them also had effects on tooth morphology.[11]

Gene or location	SNPs	Chr	Start	Stop	PGP	MGP	Description
EDAR	rs3827760	2	109513601	109513601	Lulu	Arlene	Incisor shape, mesiodistal diameters
WNT10A	rs7349332	2	219756383	219756383	Alan	Arlene	Crown size
PAX9	4	14	37126773	37147012	Alan	Arlene	Tooth size variation, shoveling

[11] Ryosuke Kimura, et al., " Common polymorphisms in WNT10A affect tooth morphology as well as hair shape," in *Human Molecular Genetics* (2015) 1-8, hmg.oxfordjournals.org, accessed July 2016; and,

WC Lee, et al., Association of common PAX9 variants with permanent tooth size variation in non-syndromic East Asian populations," in *Journal of Human Genetics* Vol 57:10 (October 2012) 654-659, www.ncbi.nlm.nih.gov, accessed July 2016; and,

Ryosuke Kimura, et al., "A common variation in EDAR is a genetic determinant of shovel-shaped incisors," in *American Journal of Human Genetics,* Vol. 85:4 (October 2009) 528-535, www.ncbi.nlm.nih.gov, accessed July 2016.

Hand Shape and Development

The development of the hand is regulated by five homeobox genes which are expressed at different times and in different combinations during embryonic development, leading to the creation of five digits on the hand.[12]

Gene or location	Chr	Start	Stop	PGP	MGP	Description
HOXD10	2	176981492	176984670	Alan	Arlene	differentiation of digits
HOXD8	2	176994422	176997423	Alan	Arlene	differentiation of digits
HOXD11	2	176971721	176976563	Alan	Arlene	differentiation of digits
HOXD1	2	177053307	177055635	Alan	Arlene	differentiation of digits
HOXD13	2	176957532	176960666	Alan	Arlene	differentiation of digits

Hair Color

Hair color is determined by pigments produced by the body called melanins. The genes that produce and regulate these pigments have strong effects on hair, eye and skin color. Using genome-wide association studies, scientists have successfully identified many mutations and genes that have strong to marginally significant effects on hair color.[13] In this case, Paul inherited the majority of hair color determining genes from Lulu and Richard.

---

[12] Clifford J. Tabin, "Why we have (only) five fingers per hand: Hox genes and the evolution of paired limbs," in *Development* vol.116 (1992) 289-296, dev.biologists.org, accessed July 2016.

[13] Kaustubh Adhikari, et al., "A genome-wide association scan in admixed Latin Americans identifies loci influencing facial and scalp hair features," in *Nature Communications,* vol. 7 (1 March 2016) www.nature.com, accessed July 2016; and,

Wojciech Branicki, et al., "Model-based prediction of human hair color using DNA variants," in *Human Genetics,* vol. 129:4 (April 2011) 443-454, link.springer.com, accessed July 2016.

Nevertheless, he did inherit one gene each from Alan and from Arlene, each of which are significant determinants of hair color.

Gene or location	SNPs	Chr	Start	Stop	PGP	MGP	Description
SLC45A2	18	5	33944721	33984780	Lulu	Arlene	Hair Color (dark)
IRF4	2	6	391739	411443	Lulu	Richard	Hair Color/ Hair Graying (black)
TYR	2	11	88911040	89028927	Lulu	Richard	Hair Color (brown)
HERC2	5	15	28356183	28567313	Lulu	Richard	Hair Color (brown and black vs. red)
SLC24A5	11	15	48413169	48434926	Lulu	Richard	Hair Color (black)
OCA2	7	15	28000021	28344483	Lulu	Richard	Hair Color (brown vs. red)
MC1R	11	16	89984287	89987385	Alan	Richard	Red hair vs. Blond
ASIP	3	20	32829188	32857150	Alan	Arlene	Red vs. dark
SLC24A4	2	14	92788925	92967825	Alan	Arlene	(blond)
KITLG	rs12821256	12	89328335	89328335	Lulu	Richard	Blond hair Europeans
TPCN2	4	11	68816350	68858072	Lulu	Richard	(blond vs. brown)
EXOC2	rs4959270	6	457748	457748	Lulu	Richard	Black
TYRP1	3	9	12693385	12710285	Lulu	Richard	Hair color

Hair Shape

Hair shape is determined by many factors, but some of the most important ones in the determination of hair shape include the shape, position, balance and distribution of composite materials of the hair follicle. Through genome-wide studies, researchers have identified some of the genes that are important for hair shape variation.[14] In this case, even though Paul inherited the majority of his hair shape determining genes from Lulu and from Arlene, he inherited the EDAR gene (the most significant determinant of hair shape) from Alan.

Gene or location	SNPs	Chr	Start	Stop	PGP	MGP	Description
TCHH	10	1	152078793	152087930	Lulu	Arlene	straight vs. curly
EDAR	Many	2	152078793	152087930	Alan	Arlene	hair thickness, straight
GATA3	2	10	8087294	8117164	Alan	Richard	hair shape
PRSS53	rs11150606	16	31099011	31099011	Lulu	Richard	curly hair
FGFR2	rs4752566	10	123267631	123267631	Alan	Arlene	hair thickness
TCHH	4	1	152078793	152087930	Lulu	Arlene	
FRAS1	rs1268789	4	79280693	79280693	Lulu	Arlene	

Balding Patterns

Male pattern balding has strong genetic underpinnings. The following genes have been identified as having significant effects on balding patterns.[15] In this case, though Paul

---

[14] Kaustubh Adhikari, et al., "A genome-wide association scan in admixed Latin Americans identifies loci influencing facial and scalp hair features," in *Nature Communications,* vol. 7 (1 March 2016) www.nature.com, accessed July 2016; and,

Gillian E Westgate, Natalia V Botchkareva and Desmond J. Tobin, "The biology of hair diversity," in *International Journal of Cosmetic Science,* Vol. 35:4 (August 2013) 329-336, onlinelibrary.wiley.com, accessed July 2016.

[15] Kaustubh Adhikari, et al., "A genome-wide association scan in admixed Latin Americans identifies loci influencing facial and scalp hair features," in *Nature Communications,* vol. 7 (1 March 2016) www.nature.com, accessed July 2016; and,

inherited the majority of his balding genes from Alan and Richard, the most significant genes affecting balding patterns were inherited from Arlene.

Gene or location	Chr	SNPs	Start	Stop	PGP	MGP	Description
GRID1	10	4	87359312	88126250	Alan	Richard	
AR	X	2	66763874	66950461	NA	Arlene	
EDA2R	X	2	65815479	65859140	NA	Arlene	
PAX1	20	2	21686297	21699124	Alan	Arlene	
FOXA2	20	2	22561642	22566101	Alan	Arlene	
HDAC9	7	2	18126572	19039135	Lulu	Richard	
C1orf127 / TARDBP	1	rs12565727	11033082	11033082	Lulu	Richard	
HDAC4	2	rs9287638	239694631	239694631	Lulu	Richard	
AUTS2	7	rs6945541	68611960	68611960	Lulu	Arlene	
SPPL2C	17	rs12373124	43924219	43924219	Alan	Richard	
SETBP1/ SLC14A2	18	rs10502861	42800148	42800148	Alan	Arlene	
SUCNR/ MBNL1	3	rs4679955	151653368	151653368	Alan	Richard	

Stefanie Heilmann-Heimbach, et al., "Hunting the genes in male-pattern alopecia: how important are they, how close are we and what will they tell us," in *Experimental Dermatology,* vol. 25:4 (April 2016) 251-257, onlinelibrary.wiley.com, accessed July 2016.

EBF1	5	rs7349332	158122916	158526788	Alan	Richard	
WNT10A	2	rs7349332	219756383	219756383	Alan	Arlene	
SSPN/ ITPR2	12	rs9668810	26426420	26426420	Lulu	Richard	

Beard Thickness

The thickness of the beard is determined by some of the same genes that influence scalp hair. Some researchers have identified the following genes and mutations as having significant effects on the density of beard growth.[16]

Gene or location	**Chr**	**SNPs**	**Start**	**Stop**	**PGP**	**MGP**	**Description**
EDAR	2	Many	109510927	109605828	Lulu	Arlene	
LNX1	4	rs4864809	54511913	54511913	Lulu	Arlene	
PREP	6	rs6901317	106036151	106036151	Lulu	Arlene	
FOXP2	7	3	113726365	114333827	Lulu	Arlene	

Skin Color

Skin color is also determined by the expression of pigments and melanin in the skin and many of the genes that influence eye and hair color also influence skin color.[17]

[16] Kaustubh Adhikari, et al., "A genome-wide association scan in admixed Latin Americans identifies loci influencing facial and scalp hair features," in *Nature Communications,* vol. 7 (1 March 2016) www.nature.com, accessed July 2016.

[17] Richard A. Sturn, "Molecular Genetics of human pigmentation diversity," in *Human Molecular Genetics,* vol. 18 (2009) R9-R17, hmg.oxfordjournals.org, accessed July 2016.

Gene or location	Chr	Start	Stop	PGP	MGP	Description
MC1R	16	89984287	89987385	Alan	Richard	
ASIP	20	32829188	32857150	Alan	Arlene	
SLC24A5	15	48413169	48434926	Lulu	Richard	
IRF4	6	391739	411443	Lulu	Richard	
OCA2	15	28000021	28344483	Lulu	Richard	
HERC2	15	28356183	28567313	Lulu	Richard	
TYR	11	88911040	89028927	Lulu	Richard	
SLC45A2	5	33944721	33984780	Lulu	Arlene	
TYRP1	9	12693385	12710285	Lulu	Richard	
KITLG	12	88886570	88974250	Lulu	Richard	
SLC24A4	14	92788925	92967825	Alan	Arlene	

Face Shape (Chin, Forehead, Width, Length, Cheeks)

Morphology of the face is determined by underlying bone, muscle, ligaments and fat and many genes have been found to have a significant effect on the shape of the face. [18]

Gene or location	Chr	Start	Stop	PGP	MGP	Description
EDAR	2	109510927	109605828	Lulu	Arlene	Chin protrusion

[18] Kaustubh Adhikari, et al., "A genome-wide association scan implicates DCHS2, RUNX2, GLI3, PAX1 and EDAR in human facial variation," in *Nature Communications,* vol. 7 (19 May 2016) www.nature.com, accessed July 2016; and,

Fan Liu, et al., "A Genome-Wide Association Study Identifies Five Loci Influencing Facial Morphology in Europeans," in *PLOS Genetics* (13 September 2012), journals.plos.org, accessed July 2016.

PAX3	2	223064606	223163715	Alan	Arlene	Distance from eyes to the nasion (nasal bridge)
TP63	3	189348942	189615068	Alan	Arlene	Distance between eyes
SMIM23	5	171212819	171218129	Alan	Arlene	Distance from zygion to the nasal bridge; distance from eyes to the nasal bridge
COL17A1	10	105791046	105845638	Alan	Arlene	Distance eyes to the nasal bridge

Height

Height is one of the most thoroughly researched genetic traits and to date more than 180 variants have been found to account for normal variation in human height. Some of the following genes have significant effects.[19]

Gene or location	Chr	Start	Stop	PGP	MGP	Description
SCMH1	1	41492871	41707815	Lulu	Richard	
IHH	2	219919142	219925238	Alan	Arlene	

[19] Michael N. Weedon and Timothy M. Frayling, " Reaching new heights: insights into the genetics of human stature," in *Trends in Genetics,* vol. 24:12 (December 2008) 595-603, www.sciencedirect.com, accessed July 2016; and,

Guillaume Lettre, "Recent progress in the study of genetics of height," in *Human Genetics,* vol. 129:5 (May 2011) 465-472, link.springer.com, accessed July 2016.

ANAPC13	3	134196546	134204865	Alan	Richard	
ZBTB38	3	141043055	141168634	Alan	Richard	
HHIP	4	145567148	145662542	Alan	Richard	
BMP6	6	7727011	7881972	Lulu	Richard	
HMGA1	6	34204577	34214008	Lulu	Richard	
PPARD	6	35310335	35395968	Lulu	Richard	
GNA12	7	2767739	2883963	Lulu	Richard	
CDK6	7	92234235	92465941	Lulu	Arlene	
PLAG1	8	57073463	57123859	Alan	Arlene	
PXMP3	8	77892494	77913280	Alan	Arlene	
PTCH1	9	98205264	98279247	Lulu	Richard	
HMGA2	12	66218240	66360071	Lulu	Richard	
SOCS2	12	93963598	93970521	Lulu	Richard	
ACAN	15	89346674	89418585	Lulu	Arlene	
RNF135	17	29296224	29326929	Alan	Arlene	
NOG	17	54671060	54672951	Alan	Richard	
TBX2	17	59477257	59486827	Alan	Richard	
DYM	18	46567846	46987172	Alan	Arlene	
BMP2	20	6748745	6760910	Alan	Richard	
GDF5	20	34021149	34026027	Alan	Arlene	
SPAG17	1	118496288	118727848	Lulu	Arlene	
rs11205277	1	149892872	149892872	Lulu	Arlene	
DNM3	1	171810618	172387606	Lulu	Arlene	
TSEN15	1	184020811	184043346	Lulu	Arlene	
ZNF678	1	227751220	227865144	Lulu	Richard	
EFEMP1	2	56093097	56151298	Lulu	Arlene	
NCAPG	4	17812436	17846488	Alan	Arlene	
rs10946808	6	26233387	26233387	Lulu	Richard	

rs2844479	6	31572956	31572956	Lulu	Richard	
rs185819	6	32050067	32050067	Lulu	Richard	
LIN28B	6	105384874	105531207	Lulu	Arlene	
C6orf173	6	126660660	126670548	Lulu	Arlene	
ADGRG6	6	142623056	142767403	Lulu	Richard	
ZNF462	9	109622469	109775525	Lulu	Richard	
DLEU7	13	51286759	51417885	Alan	Richard	
FBLN5	14	92335755	92414046	Alan	Arlene	
SH3GL3	15	84116091	84287495	Lulu	Arlene	
ADAMTS17	15	100511643	100882183	Alan	Richard	
CABLES1	18	20714528	20840434	Alan	Arlene	
DOT1L	19	2164148	2232577	Lulu	Arlene	
SUPT3H	6	44794467	45345788	Lulu	Arlene	
EXT1	8	118811602	119124058	Alan	Arlene	
FREM1	9	14734664	14910993	Lulu	Richard	
PALM2	9	112403068	112713756	Lulu	Richard	
AKAP2	9	112810878	112934792	Lulu	Richard	
NUP37	12	102467967	102514025	Lulu	Richard	
PMCH	12	102590237	102591614	Lulu	Richard	
IGF1	12	102789645	102875563	Lulu	Richard	
KRT20	17	39032141	39041495	Alan	Arlene	
ANKRD60	20	56793510	56804039	Alan	Richard	
LHX3	9	139088096	139096955	Lulu	Arlene	
QSOX2	9	139098179	139137687	Lulu	Arlene	

Conclusion

Through this project we were able to identify the segments of DNA that Paul received from each of his grandparents for 96.7% of his genome. We were also able to trace the inheritance pattern of many genes that have a physical manifestation in Paul's characteristics. During future research, we would recommend continuing to perform chromosome mapping for more distant generations by collaborating with close and distant genetic cousins and determining the origin of their shared DNA.

Our team of genealogists can also perform traditional research to discover the stories that affected Paul's life today and that of his family.

We have very much enjoyed aiding in the discovery of Paul's genetic relationship to his grandparents and look forward to performing additional research and analysis in the future.

PAW/chf

https://legacytree.com