Entire Maternal Chromosome 21 Inherited from 5th Great Grandparents – Captain Daniel and Elizabeth (Windecker) Young

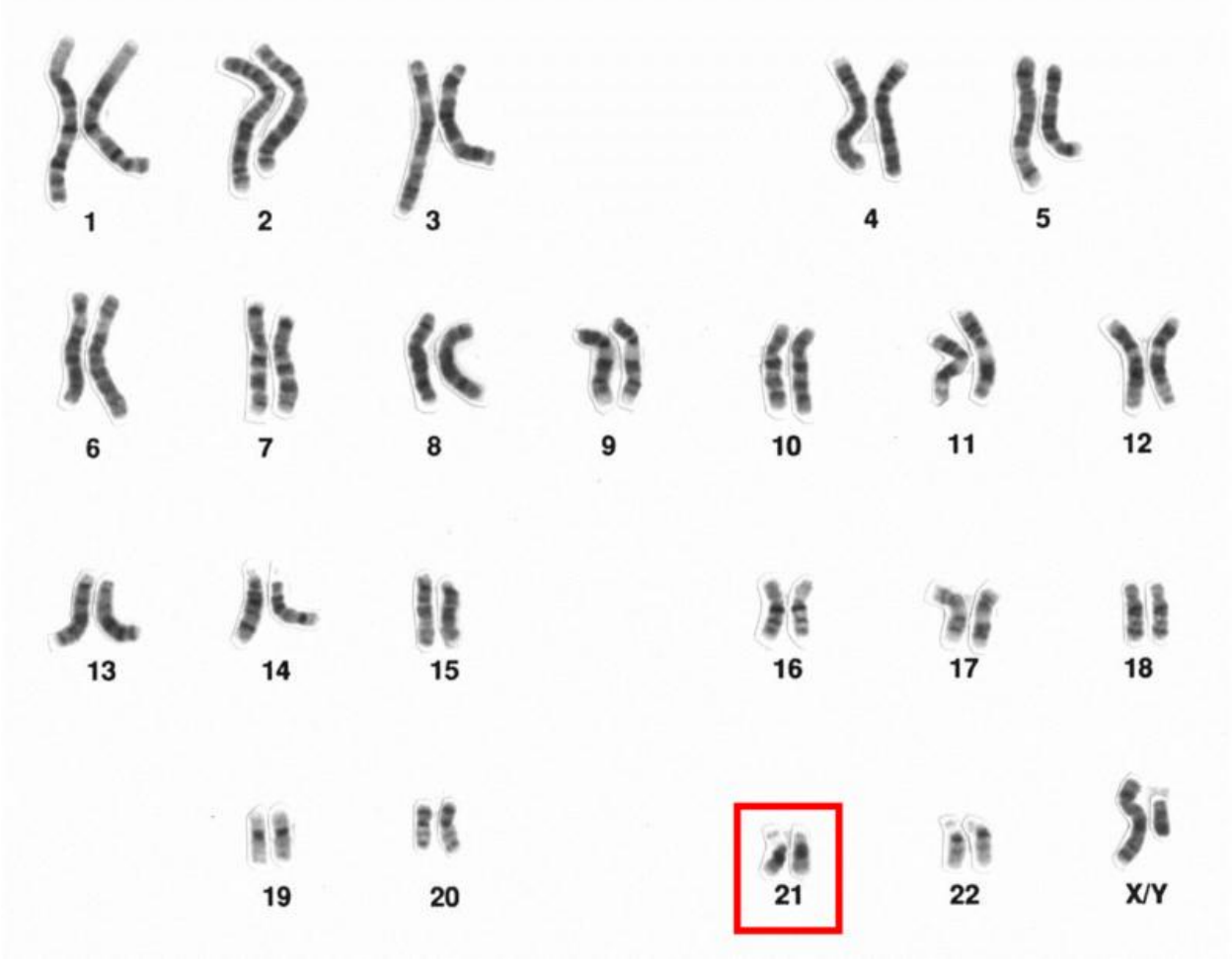
The Ancestors: Daniel Young was born about 1755 in the Canajoharie District of the Mohawk Valley New York, U.S.A. and died 9 May 1836 at his home in Barton Township (now City of Hamilton), Wentworth County, Ontario, Canada. His father was Johann Adam Jung (Young) who was born in 1717 at Foxtown, Schoharie Valley, New York, and died 1790 at the Grand River in what is today Seneca Township, Haldimand County, Ontario. Daniel's mother was Catharine Elizabeth Schremling who was born about 1720 in the Schoharie Valley, New York and died in 1798 at the home of her son Daniel in Barton Township. Daniel was a Sergeant in Butler's Rangers during the American Revolution, and a Captain of the 5th Lincoln Militia during the War of 1812. An overview of his illustrious career can be seen here. Daniel's wife was Elizabeth Windecker, born about 1763, probably on the Windecker Tract, Canajoharie District, New York, and died at her home in Barton Township on 8 March 1829. Elizabeth's father was Hendrick Windecker (a "notorious" Private in Butler's Rangers) born about 1738 probably on the Windecker Tract, and died after 1814 likely at the Grand River, North Cayuga Township, Haldimand County, Ontario. Her mother was Dorothy Pickard born about 1743 in the Canajoharie District, date and place of death unknown.

Author's Genealogical Connection: The extensive paper documentation travels back in time from David K. Faux to his mother Violet M. Williamson to her mother Eva F. Dawson to her father Joseph W. Dawson to his mother Hannah Adelia Young to her parents Henry Young and Elizabeth M. Young (first cousins). Henry's father was Henry Young Sr. and mother Rachel Young (a first cousin once removed to her husband). Elizabeth M. Young's father was George Young and her mother was Mary Terryberry. Henry Young Sr. and George Young were brothers, both the sons of the above Daniel and Elizabeth – making the latter the 5th great grandparents (twice over) to David K. Faux.

DNA Testing: The autosomal (22 chromosome pairs) DNA of David K. Faux was tested by Ancestry.com and 23andMe.com and further analyzed by Gedmatch.com. Cousins of varying degrees were also tested and analyzed by one or more of the above firms. Key matches to segments along chromosome 21 (and others) were informative cousins (close cousins share too many lineages to be certain of how to interpret the results) ranging from half third cousins once removed and 4th cousins (where a matching segment on a chromosome can be assigned to the Young / Windecker family), to 5th and 6th cousins (where the match can often be isolated to either the Young or the Windecker family) – basically those with whom the only connection, meaning ancestors in common, was via the Young or Windecker families. Thus in some instances a matching segment between two distant cousins could be deemed to be from Daniel Young or Elizabeth Windecker (and in some cases even to the level of their parents).

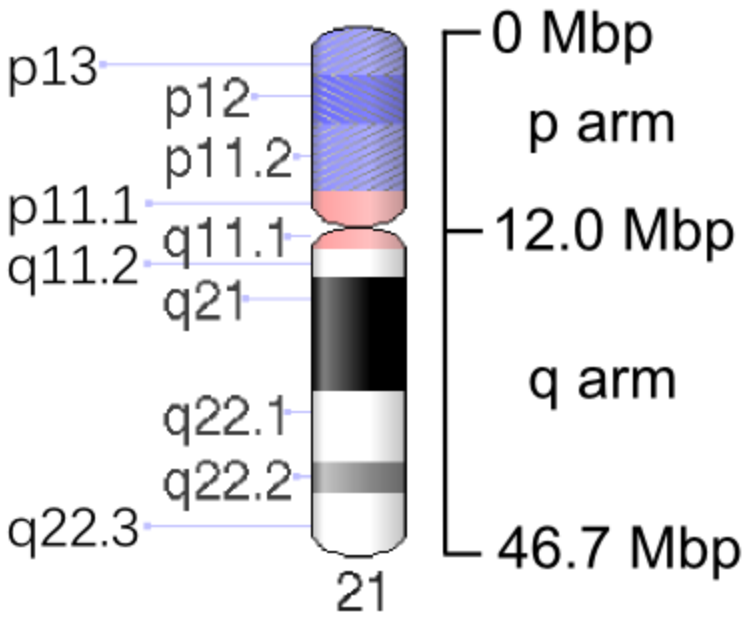
<u>An Overview of Chromosome 21</u>: Before delving into how the author is able to say with certainty that the entirety of his maternal chromosome 21 comes from his 5th great grandparents, it will be useful to provide a brief description of the nature of the this chromosome.

To visualize chromosome 21 within the context of the human genome a good starting point is a karyogram to show what it "looks like" at least at one stage of the process of cell division. If for example a blood sample is drawn and say a white blood cell is "opened up" to view the nucleus and its contents when during cell division the chromosomes have isolated themselves into discrete entities during division, what follows is what one would see:



It can be seen that chromosome 21 is the smallest of the 22 autosomes (the 23rd pair being the sex chromosomes), and that there are two – one from the father and the other from the mother.

Another useful way of visualizing chromosome 21 is diagrammatically where the different regions can be displayed, and their distinguishing features noted, as seen below:



G-bands of human chromosome 21 in resolution 850 bphs[17]

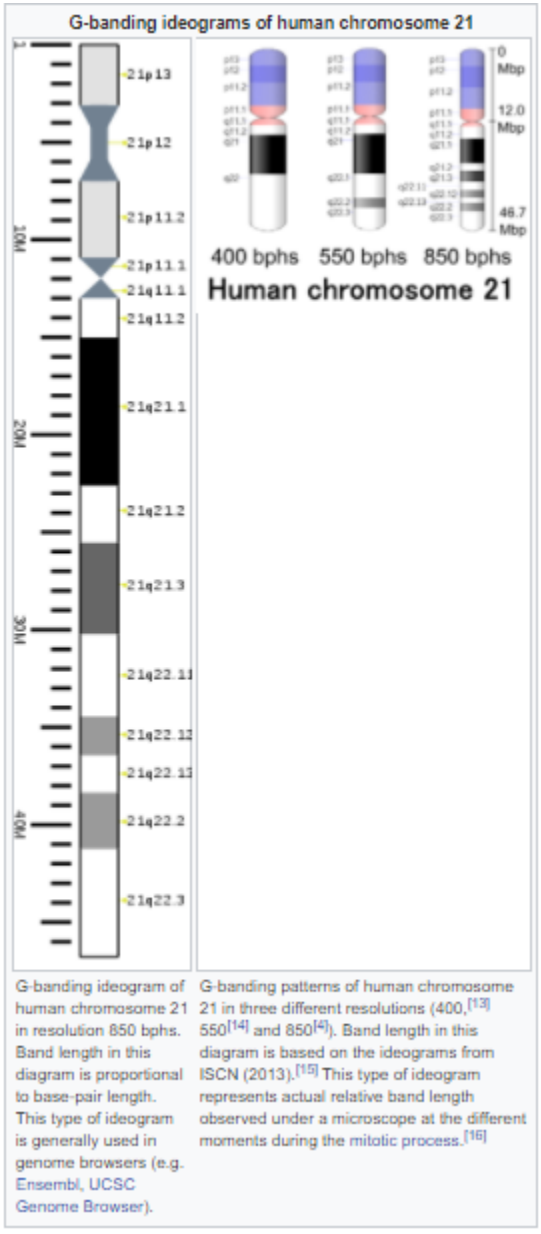
Chr.	Arm[18]	Band[19]	ISCN start[20]	ISCN stop[20]	Basepair start	Basepair stop	Stain[21]	Density
21	p	13	0	311	1	3,100,000	gvar	
21	p	12	311	683	3,100,001	7,000,000	stalk	
21	p	11.2	683	1056	7,000,001	10,900,000	gvar	
21	p	11.1	1056	1274	10,900,001	12,000,000	acen	
21	q	11.1	1274	1367	12,000,001	13,000,000	acen	
21	q	11.2	1367	1584	13,000,001	15,000,000	gneg	

21	q	21.1	1584	2019	15,000,001	22,600,000	gpos	100
21	q	21.2	2019	2144	22,600,001	25,500,000	gneg	
21	q	21.3	2144	2330	25,500,001	30,200,000	gpos	75
21	q	22.11	2330	2485	30,200,001	34,400,000	gneg	
21	q	22.12	2485	2610	34,400,001	36,400,000	gpos	50
21	q	22.13	2610	2703	36,400,001	38,300,000	gneg	
21	q	22.2	2703	2858	38,300,001	41,200,000	gpos	50
21	q	22.3	2858	3200	41,200,001	46,709,983	gneg	

The length of the chromosome in total is 46.7 Mb, meaning 46,700,000 base pairs in length (other work gives as much as 48,129,895 bp. So you may have say an A nucleotide base (a SNP or single nucleotide polymorphism) from your mother and a C from your father at one location along the chromosome – the total being over 46 million pairs of them. For our purposes this is what is most important due to what is being tested, but it is noteworthy that a chromosome included many other elements such as short tandem repeats (STRs) such as strings of say ACGACGACG, insertions, deletions and perhaps an inversion, and a host of other characteristics that do not come into play for our purposes. Although Hattori found only 127 known genes, 98 predicted genes and 59 pseudogenes, more recent research suggests that the number may be as high as between 477 and 635 genes. The above ideogram does not show all the bands that appear on chromosome 21 after staining and viewing under a microscope – each band being used to help define the location of a gene.

The chromosome is composed of euchromatin (the q arm) which is what is being tested by the various commercial companies, and heterochromotin (the p arm) which is a complex tangle shown in blue above that contains few if any genes and which is lacking in many identifyable SNPs. The pink section above is the centromere, separating the p (short) and q (long) arms, and

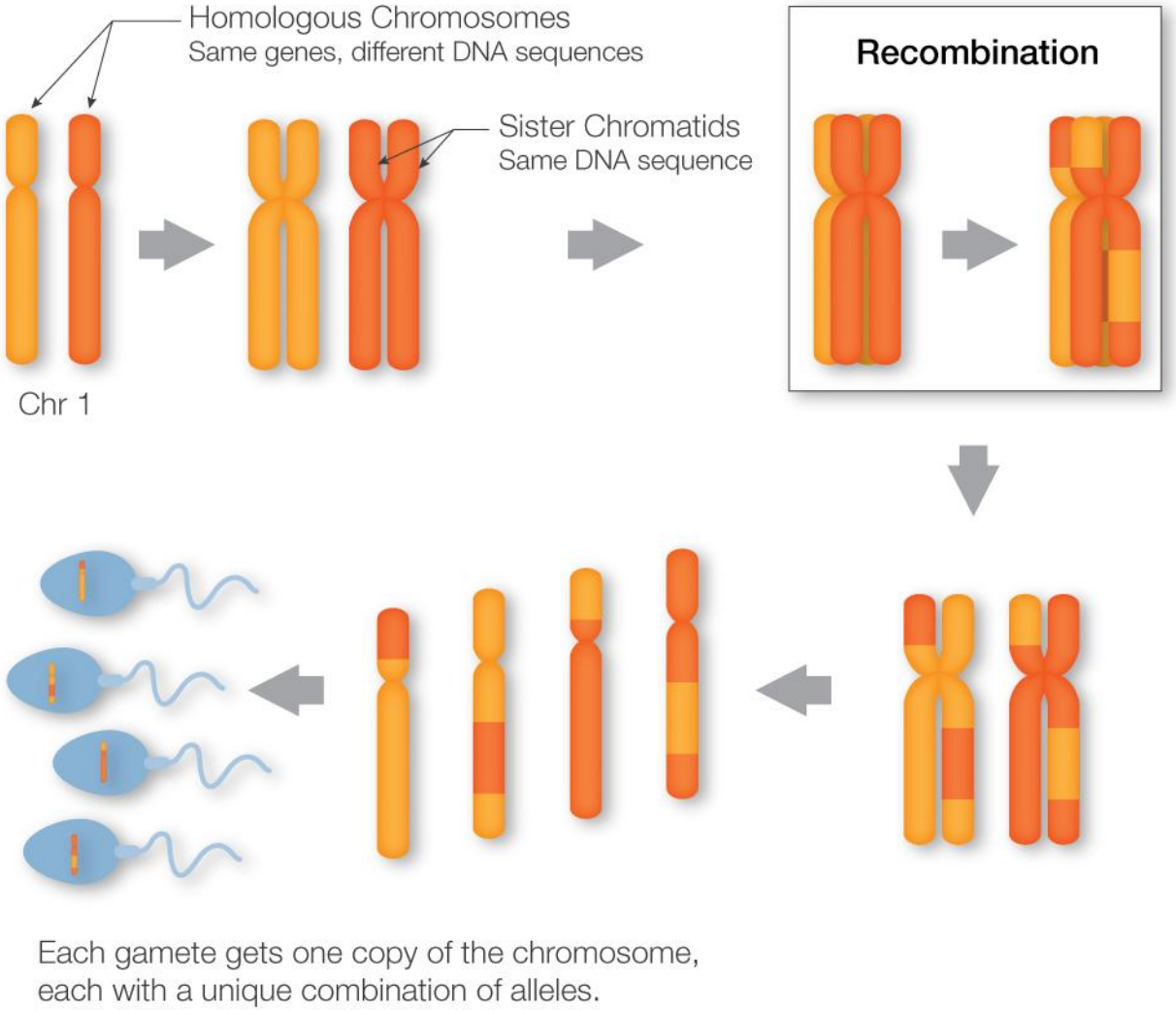
which is also very SNP poor and largely uninformative in terms of ancestry and family matching, and its origin must to some extent be inferred. Thus for all intents and purposes what can be said definitively relates to the q arm alone, and only the SNPs from about 14 Mb shown at the top of the dark black stained band known as q21 to the telomere (end section) of the distal end at 46.7 Mb.



Some of the "performance characteristics", particularly in terms of what happens in meiosis (the formation of eggs and sperm) of chromosome 21 have likely impacted the origin of the segments found here in the author. One key finding is that there is considerably less male

recombination than female recombination during meiosis (in the order of 1.6 female recombinations to every 1 male recombination). This means that on average male recombinations will produce longer segments, and female recombinations more but shorter segments – "breaking up" the chromosome more.

During meiosis chromosomes must go from the diploid (possessing 2 of each chromosome) to the haploid (possessing one of each chromosome) state. The X shaped chromosome joined at the centromere lines up with the sister chromatid from the mother during egg production, and the same process occurs in the father during sperm production, then the process of recombination occurs. To picture recombination, the following diagram should help:

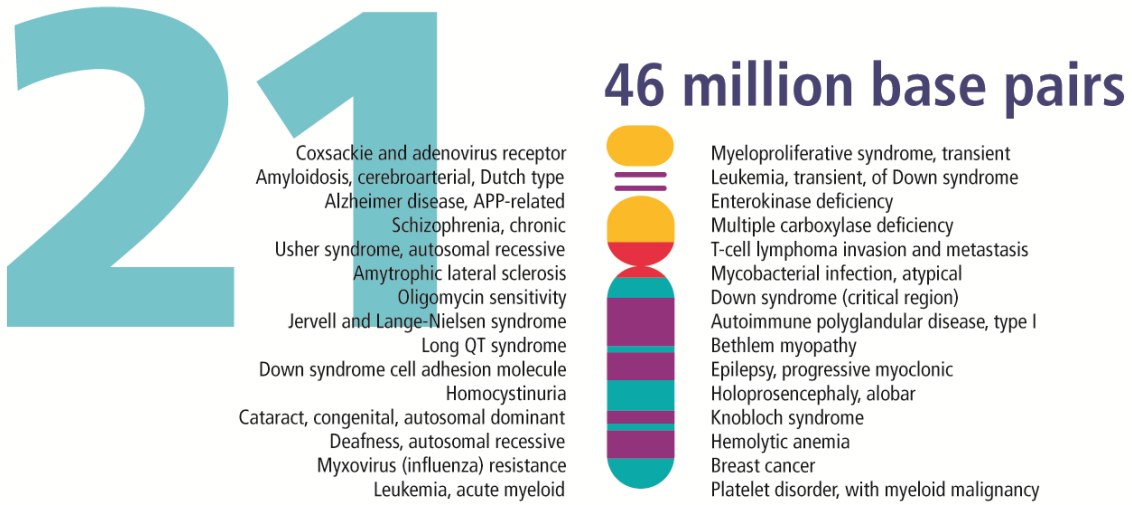


Here, during the process of recombination, the chromatids exchange genetic material at a chiasma (point for disjunction or break apart and junction or splicing) so that each new pair will have chunks (blocks or segments) from its homologous mate, meaning that in a female the chromosome that gravitates to one of the four eggs (gametes) formed after division has perhaps

two new segments sliced from the other chromosome in the pair. During fertilization one of the chromatids from the mother will merge with the contents of the gamete of the father. Then cell division from this point is called mitosis. At a certain phase of mitosis (division of body cells to form a duplicate), for example white blood cells, it is possible to isolate each individual chromosome from the mother and father as seen in the karyogram above. Hence the new chromosome compliment is a combination of the two original units, one from the mother and one from the father.

The randomness and sheer number of possibilities of what emerges in the formation of the four gametes above is staggering. However one of the possibilities is that an entire chromosome can be passed to the next generation – but the likelihood of this happening such that someone obtains an entire maternal or paternal chromosome from 5th great grandparents is vanishingly small since there are 6 meiotic recombination events or opportunities for segments to be spliced from the other 30 maternal ancestors. It would mean that for all intents and purposes there was no recombination, effectively it did not happen at all, for the events leading up to what is inherited by a 5th great grandchild. In the author's case, the outcome was to some degree assisted by a cousin marriage, but this would explain only a small fraction of the whole picture.

Diseases Associated with Chromosome 21:



Genes Associated with Chromosome 21:

The following are some of the approximately 215 genes located on chromosome 21:

- APP: amyloid beta (A4) precursor protein (peptidase nexin-II, Alzheimer disease)[11]
- C21orf45: encoding protein Protein Mis18-alpha
- C21orf55/DNAJC28: encoding protein DnaJ homolog subfamily C member 28

- C21orf56: encoding protein Uncharacterized protein C21orf56
- C21orf59: Chromosome 21 open reading frame 59
- C21orf62: expressed in tissues of the brain and reproductive organs
- C21orf66: encoding protein GC-rich sequence DNA-binding factor homolog
- CBS: cystathionine-beta-synthase
- CLDN14: claudin 14
- CRYZL1: Crystallin zeta-like 1
- CYYR1: Cysteine and tyrosine rich 1
- DIP2A: Disco-interacting protein 2 homolog A
- DOPEY2: Dopey family member 2
- DSCR1: Down Syndrome critical region 1[12]
- DYRK1A: dual specificity tyrosine-(Y)-phosphorylation regulated kinase 1A
- FAM3B: Family with sequence similarity 3 member B
- FRGCA: encoding protein FOXM1-regulated, gastric cancer associated
- HLCS: holocarboxylase synthetase (biotin-(propionyl-Coenzyme A-carboxylase (ATP-hydrolysing)) ligase)
- KCNE1: potassium voltage-gated channel, Isk-related family, member 1
- KCNE2: potassium voltage-gated channel, Isk-related family, member 2
- LAD: leukocyte adhesion deficiency (symbols are ITGB2, CD18, LCAMB)
- PCNT: centrosomal pericentrin
- PDXK: encoding enzyme Pyridoxal kinase
- PSMG1: Proteasome assembly chaperone 1
- RNR4: RNA, ribosomal 45S cluster 4
- RRP1: encoding protein Ribosomal RNA processing protein 1 homolog A
- RRP1B: ribosomal RNA processing 1 homolog B
- RWDD2B: encoding protein RWD domain-containing protein 2B
- S100B: calcium binding protein
- SOD1: superoxide dismutase 1, soluble (amyotrophic lateral sclerosis 1 (adult))
- TMEM1: encoding protein Trafficking protein particle complex subunit 10
- TMPRSS3: transmembrane protease, serine 3

- TTC3: Tetratricopeptide Repeat Domain 3

Evidence that Chromosome 21 was Inherited Entirely from Daniel and Elizabeth (Windecker) Young:

The evidence comes from the matching segments of individuals with a known genealogy. Most of these can be displayed in the match profile from Gedmatch.com from individuals who uploaded their raw data to that site, and comparing this to their documented genealogy. This is seen in the diagram below, using the phased maternal data of the author:

T384570	21	9,849,404	16,699,698	8.7	740	Ferne Bolton	F	fauxdk@yahoo.com	
M812348	21	16,658,736	26,769,135	18.7	2,801	Norm Sones	U	nelson.tom@sympatico.ca	
A974457	21	16,658,736	19,341,200	7.1	798	*Marti Sigsbee	F	cheryl6b@yahoo.com	
M121610	21	16,969,464	43,652,459	54.3	7,962	K.L.	M	fauxdk@yahoo.com	
A974287	21	18,330,173	22,749,282	8.7	1,284	Robert Lloyd Hall	U	bhall@mill-ore.com	
A247392	21	18,344,736	38,231,969	33.9	5,380	Lawrence William Brown	M	larrook@yahoo.com	
T794047	21	20,915,614	38,276,905	28.4	4,705	*Kathryn Willson	F	kathrynparchelo@gmail.com	
A668294	21	21,271,294	27,141,178	9.0	1,002	Gail Crichton	F	oxtower2@aol.com	
A009834	21	23,901,585	33,066,196	13.7	2,359	Nola Helen King	U	sharon.swonger@yahoo.com	
A048311	21	23,901,585	30,509,664	9.6	1,120	*RobertsMaiden	F	MiMi.5695@gmail.com	
A582381	21	24,060,742	33,090,443	13.4	1,477	*sefp	F	sefpatrick@yahoo.com	
A862071	21	25,012,337	37,004,682	19.8	2,031	Jocelyn Malheiro	F	jocelyn.malheiro@gmail.com	
A442476	21	26,798,079	31,793,748	7.0	1,293	Barry Shumaker	M	birddog23@sasktel.net	
T384570	21	27,311,934	46,909,175	41.2	6,108	Ferne Bolton	F	fauxdk@yahoo.com	
A560066	21	29,944,093	36,342,520	11.0	1,730	Anthony Messuri Jr	M	rpgpgmr@mindspring.com	
A586493	21	30,683,684	38,222,301	14.0	1,321	Rashelle Elburg	F	Rashellenewman@hotmail.com	
M812348	21	38,145,277	45,159,710	19.4	2,616	Norm Sones	U	nelson.tom@sympatico.ca	

The chart names the chromosome, the start and stop matching segment in Mb, the cM (genetic size of the match), and the number of matching SNPs (single nucleotide polymorphisms). Note that a cM is a measure of genetic distance whereas Mb is a measure of physical distance. As a very general rule, one cM = 1 Mb, but there are notable exceptions.

As to the known individuals in the chart above (and others):

The p end of the chromosome is shown by the match with **Ferne Bolton**, the author's sister. It has so few SNPs (markers) that anything to somewhere between 14 and 16 Mb is not particularly informative. This means that it is not possible to say with complete certainty anything about the p end either that here it is from the maternal side. Ferne also shares the last half of the chromosome, with the intervening part shared with a first cousin (and thus a Williamson segment).

Norm Sones is a half 3rd cousin once removed with the ancestor in common being the author's ggg grandmother Elizabeth M. Young. He matches in two locations on this chromosome – the distal ends.

K.L. is the author's second cousin. Our maternal grandmothers were sisters. It appears that K.L. shares almost as much of chromosome 21 as the author, with the exception of small amounts at each end.

Lawrence William Brown is a 5th cousin once removed, a descendant of Peter Young (via his daughter Catharine Cramer), brother to the author's 4th great grandfathers, Henry Young and George Young.

Bob Hall is also a 5th cousin once removed, and a descendant of Peter Young but via his son Edmund Wellington Young. Bob's data is not on Gedmatch at this point but our match was from a start point of 18,330,173 to 22,749,282. The match is 8.7 cM.

Kathryn Willson is a 5th cousin twice removed, a descendant of Barbara Windecker (who married James Fleming), daughter of Henry Windecker (father of Elizabeth who married Daniel Young – the parents of Henry, George, Peter and others) and Dorothy Pickard.

Jocelyn Malheiro and the author's closest connection is via Hans Bellinger who married Anna Bellinger in 1642, Jocelyn being a descendant of son Dietrich Bellinger born 1644 and the author a descendant of son Nicholas Bellinger born 1645. This makes us 11th cousins! None the less, there are no other ancestors known to be shared in common – and the author has similar relatively large matches to descendants of distant ancestors in other lines. This is a Windecker connection since Henry Windecker's great grandfather was Nicholas Bellinger.

Barry Schumaker is a 6th cousin once removed, a descendant of Margaret Windecker (who married Stephen Kitson) daughter of Henry Windecker, sister to Elizabeth (Young) Windecker. Here also there is no connection to the Young family, so the match is Windecker or Pickard.

Anthony Messuri Jr. is a distant cousin, a descendant of Elizabeth Windecker's great aunt Gertrude Windecker (8th cousin once removed) who married Jacob Pickard (7th cousin once removed) the uncle of Elizabeth.

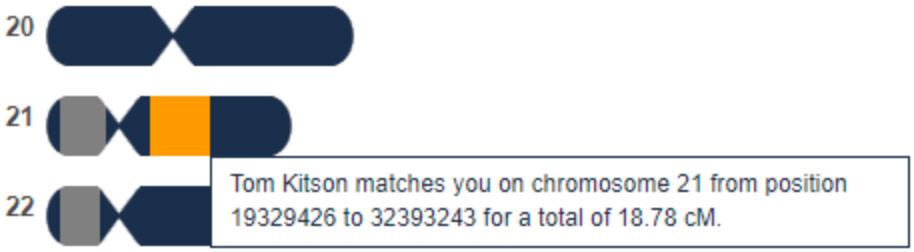
Rashelle Elburg is a known descendant of Daniel and Elizabeth (Windecker) Young.

The other individuals shown in the chart above (of which there are not many) are either undocumented Young – Windecker descendants, or are simply false positives.

Using the chart above it is clear that there are some rather astounding matches to distant kin, but that the fit on chromosome 21 makes perfect sense. One further cousin match relating to a match on this chromosome is found at 23andMe.com as follows:

Suzanne Longo, who is a 3rd cousin once removed but whose data is not uploaded to Gedmatch, matches between 40 Mb and 46 Mb. A closer examination of the data shows that the numbers are actually 40.4 Mb and 46.2 Mb. The match is 14.6 cM, and takes known the Young – Windecker segment to the q end telomere, offering further proof that the entire intact maternal chromosome is indeed from this lineage. The shared segment ends specifically at 46,263,539 for the author, Norm Sones, and Suzanne Longo. Academic research shows that the end of the chromosome is 46,709,983 (each testing company differs to some degree). So for all intents and purposes our match extends to the far q end of chromosome 21.

Tom Kitson, who is a descendant of Margaret Windecker, daughter of Henry Windecker and Dorothy Pickard who married John Kitson (Kitzer). Diagram from Family Tree DNA browser:



Some interesting Facts About Chromosome 21:

1) It is the smallest of the 22 autosomes.

2) The chromosome was first sequenced and fully described in 2000 by Hattori et al.

3) Female recombination is about 1.5 times greater on this chromosome than it is in males, with a tendency to see more near the centromere (8 times more likely) and decreases along the q arm. Male recombination in the telomeric regions is about 1.8 times that seen in females. Chromosome 21 has an overall recombination rate double the human genomic average. In both sexes there is a "deficiency" in zero exchanges [the author's family clearly being an exception], and an excess in single exchanges, with males showing far fewer multiple exchanges than females. Also there is a decrease in recombination with increasing maternal age [the author's great-great grandmother was 18 years old when she had his great-grandfather – so evidence of exceptions in some recombinations], but paternal age is not a "major determinant" of recombination in human chromosome 21q. Females and males show 63% and 25% respectively, of all meiotic exchanges in the first half of 21q. This study was by Lynn et al., 2000.

4) The centromere exerts a negative effect on recombination both within itself and in proximal regions. However, the exception as noted in the Lynn study is in females where the recombination rate is higher in the most proximal region (2.4 Mb) of the q arm to the centromere than along the rest of the arm. In females there is also a "recombinationally hyperactive region" localized at 5 Mb from the centromere (a "hot spot"). Little is known of the recombination activity along the short arm – 21p. Study by Laurent et al., 2003.

5) A series of recombination jungles (hot spots), as well as "cold spots" or deserts, have been profiled for all chromosomes by Chowhurdry et al., 2009. This relates to the q arm, from about 14.5 Mb (the effective start point for the assessment of this chromosome).

6) Chromosome 21 represents between 1.5 and 2% of the total cellular DNA – so in this one "package" the author received about 2% of his DNA.

7) Recent studies show that it includes over 450 genes, almost all on the q (long) arm.

8) This chromosome is the cause of Down's syndrome, Trisomy 21, there being three chromosomes rather than the normal two. Thus the excess proteins or other factors due to this

irregularity is what causes the features characteristic to Down's including the distinctive facial features, intellectual deficits, short fingers, thick tongue – to name a few.

9) It is associated with a variety of health conditions including Alzheimer disease, amyotrophic lateral sclerosis, familial atrial fibrillation, prostate cancer, familial combined hyperlipidemia, and predisposition to bipolar disorder to name but a few.

10) In the blog by Kitty Cooper she has a posting entitled, "Using the Chromosome Mapper to make a four generation inheritance picture. Using this tool "Byrnne" had four generations of her family tested, meaning all 8 of her great grandparents. One observation is that it was, "*Interesting that she has chromosome 21 intact from her paternal great-grandmother*". The author did not have this luxury / blessing of testing even grandparents, but the data still shows clearly (by a more circuitous route) that his maternal great great-grandmother was the source of the author's maternal chromosome 21.

11) More information can be found in the Wikipedia article for Chromosome 21, and "Genetics Home Reference" Chromosome 21 articles online.

Conclusions:

Combining all of the data above, it can be seen that there are no gaps, the author has inherited the entire maternal chromosome 21 from Daniel and Elizabeth (Windecker) Young.

It is often difficult if not impossible to parse out the contributions of the husband (Daniel) and wife (Elizabeth), but in this case for at least one segment we can be certain that Elizabeth (Windecker) Young was the one who contributed the span from 21,002,155 to 38,077,824 Mb.

It would appear that of the two sons of Daniel and Elizabeth, George Young provided the beginning and end (telomere sections) of the chromosome, and likely his brother Henry Young contributed the mid section, that is shared with Lawrence William Brown (start 18,344,173 to 38,239,633).

The entire weight of data poses an interesting question. The evidence shows that this chromosome does not come from the Pickard line (wife of Henry Windecker). Since both Henry Young Sr. and George Young were Windecker descendants and therefore Bellinger descendants, and considering the relatively large Bellinger match to an 11th cousin, it can be asked whether the entire chromosome came down intact from the early 1600s (through many potential recombination possibilities) via **Hans Bellinger** born about 1618 and his wife **Anna** Bellinger of Steinau an der Strasse, Hessen, Germany.

At one level there seems to be some sort of "stickiness" factor at work, where the chromosome had 12 chances to recombine (one recombination per generation is typical). However there does not seem to be scientific validation for such an event. Hence it is more likely that the correct interpretation is that what we are seeing is merely a very low probability occurrence in multiple

generations of one family manifesting itself - although a similar occurrence (but on a more restricted scale) is found in the large number of those from various branches of the family who share a segment of chromosome 2 which can be traced back to the 1600s via the Young or Schremling families.

There may, however, have been one recombination event that occurred but it is "disguised" to some extent due to a first cousin marriage. It seems that the author's great great grandmother, Hannah Adelia (Young) Dawson, may have received the larger mid section from her father Henry Young Jr. (and likely this part came down via the Windecker – Bellingers); whereas the smaller distal segments of chromosome 21, the parts that match Norm Sones, came from her mother Elizabeth (Young) Young (the ancestor Norm shares in common with the author). Since some Young descendants such as Lawrence William Brown entirely match the left side of the match with Norm Sones, this is a Young or Windecker segment. What is not entirely clear at present is whether the smaller segment at the distal q end matching Norm Sones and Suzanne Longo came from Elizabeth's father George Young, or her mother Mary Terryberry. There are as yet no matches that could settle this one outstanding matter. Hopefully in due course the key DNA match will emerge to verify whether the above distal match to Norm Sones and Suzanne Longo is via our Young or Terryberry ancestors.

A Question for Consideration:

The question presently on my mind is which of my grandchildren will have nothing of my maternal chromosome 21, how many will have some part, and whether perchance one or more will have the entire package. As noted, it appears that the author's second cousin K.L. also inherited most or all of chromosome 21 from the same source.

Dr. David K. Faux

Cypress, California and Caledonia, Ontario. Version: 21 October 2015. Updated 22 June 2016, 19 July 2017, and 15 November 2017.