CHROMOSOME 2: Mapping the Transitions from YOUNG / WINDECKER to BAIN and Return to YOUNG / WINDECKER Ancestral Segments

Description of Chromosome 2:

Chromosome 2 is one of the 22 pairs of autosomes (one from the mother, one from the father) and is the second largest of the group. It is characterized as, "spanning more than 242 million <u>base pairs^[5]</u> (the building material of <u>DNA</u>) and representing almost 8% of the total DNA in human <u>cells</u>." This chromosome has about 1,200 protein coding genes, including the HOXD complex, on the segment 176,880,740 to 177,056,400 Mb which codes anterior to posterior development (body structures during development) of the organism.



Above is a karyogram of the 22 pairs of human autosomes and the sex chromosomes (in a male). The size of chromosome 2 can be seen in relation to the entire array.



G-banding ideogram of human chromosome 2 ii in resolution 850 bphs. a Band length in this b diagram is proportional (to base-pair length. a This type of ideogram is generally used in t genome browsers (e.g. Ensembl, UCSC Genome Browser).	G-banding patterns of human chromosome 2 in three different resolutions (400, ^[20] 550 ^[21] and 850 ^[4]). Band length in this diagram is based on the ideograms from ISCN (2013). ^[22] This type of ideogram represents actual relative band length observed under a microscope at the different moments during the mitotic process. ^[23]
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The G banding shown in the above map allows geneticists to pin a particular gene to a physical location on the chromosome. As seen in the chart below, each band has a start and stop point expressed in Mb. Since it is not possible, for the sake of space and other factors to list every gene, the author will note examples of segments of known origin in relation to the G bands, and then show the various diseases associated with these and all bands along the chromosome.

Chr.	Arm ^[24]	Band ^[25]	ISCN start ^[26]	ISCN stop ^[26]	Basepair start	Basepair stop	Stain ^[27]	Density
2	р	25.3	0	388	1	440,0000	gneg	
2	р	25.2	388	566	4,400,001	6,900,000	gpos	50
2	р	25.1	566	954	6,900,001	12,000,000	gneg	
2	р	24.3	954	1193	12,000,001	16,500,000	gpos	75
2	р	24.2	1193	1312	16,500,001	19,000,000	gneg	
2	р	24.1	1312	1565	19,000,001	23,800,000	gpos	75
2	р	23.3	1565	1789	23,800,001	27,700,000	gneg	
2	р	23.2	1789	1908	27,700,001	29,800,000	gpos	25
2	р	23.1	1908	2027	29,800,001	31,800,000	gneg	
2	р	22.3	2027	2296	31,800,001	36,300,000	gpos	75
2	р	22.2	2296	2415	36,300,001	38,300,000	gneg	
2	р	22.1	2415	2609	38,300,001	41,500,000	gpos	50
2	р	21	2609	2966	41,500,001	47,500,000	gneg	
2	р	16.3	2966	3220	47,500,001	52,600,000	gpos	100
2	р	16.2	3220	3294	52,600,001	54,700,000	gneg	
2	р	16.1	3294	3548	54,700,001	61,000,000	gpos	100
2	р	15	3548	3757	61,000,001	63,900,000	gneg	
2	р	14	3757	3935	63,900,001	68,400,000	gpos	50
2	р	13.3	3935	4114	68,400,001	71,300,000	gneg	
2	р	13.2	4114	4248	71,300,001	73,300,000	gpos	50
2	р	13.1	4248	4353	73,300,001	74,800,000	gneg	
2	р	12	4353	4860	74,800,001	83,100,000	gpos	100

G-bands of human chromosome 2 in resolution 850 $bphs^{\rm [4]}$

Genes and Diseases Associated With Chromosome 2:

Melanoma-associated gene hyroid iodine peroxidase deficiency Goiter, congenital Hypothyroidism, congenital emia, hypobeta, abeta-, hyperbeta-, and apo-ACTH deficiency adrenal insufficiency, and red hair LCHAD deficiency Trifunctional protein deficiency, type 1 HELLP syndrome, maternal, of pregnancy Fatty liver, acute, of pregnancy Deafness, autosomal recessive Glaucoma, primary infantile Spastic paraplegia Gingival fibromatosis, hereditary Holoprosencephaly Ovarian dysgenesis Carney complexes Endometrial carcinoma Zellweger syndrome Adrenoleukodystrophy, neonatal Alstrom syndrome Preeclampsia/eclampsia Welander distal myopathy Kappa light chain deficiency Pancreatic stone protein Lissencephaly Renal tubular acidosis with deafness BRCA1-associated RING domain (breast cancer) Achromatopsia Rhabdomyosarcoma, down-regulated in Diazepam-binding inhibitor Thrombophilia due to protein C deficiency Purpura fulminans, neonatal Liver cancer oncogene Xeroderma pigmentosum, group B Trichothiodystrophy Nemaline myopathy, autosomal recessive Convulsions, familial febrile Progressive intrahepatic cholestasis Edstrom myopathy Mesomelic dysplasia, Kantaputra type Cardiomyopathy, familial hypertrophic Bardet-Biedl syndrome Ehlers-Danlos syndromes Aneurysm, familial arterial Diabetes mellitus, insulin-dependent Primary pulmonary hypertension (familial primary) Cleft palate, isolated Wrinkly skin syndrome Amyotrophic lateral sclerosis, juvenile recessive Lactic acidosis due to defect in iron-sulfur cluster of complex I Ichthyosis Finnish lethal neonatal metabolic syndrome T-cell leukemia or lymphoma Bjornstad syndrome (pili torti and deafness) Myopathy, desmin-related, cardioxeletal Cardiomyopathy, dilated Natural resistance-associated macrophage protein Hyperoxaluria, primary, type 1 Alport syndrome, autosomal recessive Hematuria, familial benign Brachydactyly-mental retardation syndrome Oguchi disease Epidermolysis bullosa

243 million base pairs

Tremor, familial essential Oculodigitoesophagoduodenal syndrome Anaplastic lymphoma kinase (Ki-1) Pseudovaginal perineoscrotal hypospadias Xanthinuria, type I Colorectal cancer, hereditary, nonpolyposis, type 1 Ovarian cancer Muir-Torre syndrome Human T-cell leukemia virus enhancer factor Precocious puberty, male Pseudohermaphroditism, male, with Leydig cell hypoplasia Hypogonadotropic hypogonadism Micropenis Leydig cell adenoma, with precocious puberty Sitosterolemia Cystinuria Doyne honeycomb degeneration of retina Dyslexia, specific Muscular dystrophy Miyoshi myopathy Myopathy, distal, with anterior tibial onset Orofacial cleft Urotacia cieft Parkinson disease, type 3 Vitamin K-dependent coagulation defect Pancreatitis-associated protein Pulmonary alveolar proteinosis, congenital Glaucoma, open angle, 8 (adult-onset) Diabetes mellitus, non-insulin-dependent Ectedermal directoria: nutreenal dominant Ectodermal dysplasia, autosomal dominant and recessive Hypothyroidism, congenital Nephronophthisis Colorectal cancer Cardiomyopathy, dilated Spastic cerebral palsy, symmetric, autosomal recessive Epilepsy Ataxia, episodic Deafness, autosomal dominant Myasthenic syndrome, slow-channel congenital Rhizomelic chondrodysplasia punctata, type 3 Cardiomyopathy, dilated Duane retraction syndrome Synpolydactyly, type II Colorectal cancer, hereditary nonpolyposis, type 3 Neurogenic differentiation Arrhythmogenic right ventricular dysplasia Myasthenia gravis, neonatal transient Cataracts Cataracts Paroxysmal nonkinesiogenic dyskinesia Choreoathetosis, familial paroxysmal Cerebrotendinous xanthomatosis AcvI-Coenzyme A dehydrogenase Carbamoylphosphate synthetase Waardenburg syndrome, types I and III Rhabdomyosarcoma, alveolar Craniofacial-deafness-hand syndrome Brachydactyly, type A1 Goodpasture antigen Serotonin receptor Bethlem myopathy Programmed cell death Leigh syndrome, French-Canadian type Ultraviolet damage, repair of Crigler-Najjar syndrome, type I

Genetic Features of Segments of Known Origin:

There are few genes of any consequence on the interesting (genealogically as will be seen below) segment between 33 to 44 Mb and 44 to 53 Mb from WINDECKER (specifically Henry WINDECKER's grandparents, Heinrick Conrad and Anna Christina (MATHEUS) WALRATH); and YOUNG ancestors (specifically Daniel YOUNG' parents Johann Adam and Catharina Elizabeth (SCHREMLING) JUNG) respectively. There are more genes on the adjacent segment from about 53 to 79 Mb which can be traced to ancestors of Mary BAIN, the source of this segment being her ancestors William SNADON (born 1700) and / or wife Catherine SNADON (born 1700), who were first cousins, from Alloa, Clackmananshire, Scotland.

Genes on the longest segment of known provenance between about 80 and 128 Mb are known to be from the author's 5^{th} great grandparents Captain Daniel YOUNG (1755 – 1834) and / or his wife Elizabeth WINDECKER (1763 – 1836). Another segment, extending from about 204 Mb to 215 Mb can be attributed to the grandparents of Daniel YOUNG, namely Johann Theobald (born 1691) and / or Maria Catharina (SNYDER) JUNG. The specifics of all segments of known origin will be depicted below in chart form after the following illustrations of the regions involved.

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<u>Chromosome</u> <u>Position (MD)</u>			<u>cM</u>	SNPs	Person	Ancestor(s) in Common	G G Grandparent(s)	Position of Match
2	8,674	6,753,810	13.2	2,128	Jim Arnold	William McCormack (1836) & Mary Bain	William McCormack & Mary Bain	
2	7,980,572	10,579,817	6.5	785	John Williams	George Dawson (1827) & Mary Ann Dunham	John Dawson	FTDNA
2	33,476,810	44,133,126	12.8	2,992	Jacqueline Adams	Henry Windecker (1736) & Dorothy Pickard	Hannah Adelia Young	
2	33,589,785	44,112,034	11.7	3,100	Jody Hash Crowl	Hendrick Conrad Walrath & Anna Christina Matheus	Hannah Adelia Young	FTDNA
2	33,589,785	44,112,034	12.1	3,200	John Bowers	Henry Windecker	Hannah Adelia Young	FTDNA

Genealogical Origins of Segments on Chromosome 2 – From Gedmatch and FTDNA:

						(1736) & Dorothy Pickard		
2	43,391,215	53,322,336	11.25	3,300	Maggie Cvek	Hendrick Schremling – Theobald Jung Sr.	Hannah Adelia Young	FTDNA
2	43,893,905	53,560,407	9.9	1,507	Robert C. Nelson	Elizabeth Young (1827)	Hannah Adelia Young	
2	43,893,905	53,462,867	9.8	1,494	Amanda Young	Daniel Young (1755) & Eliz Windecker	Hannah Adelia Young	
2	43,893,905	53,384,688	9.7	1,479	Tom Nelson	Elizabeth Young (1827)	Hannah Adelia Young	
2	43,898,184	53,384,287	9.7	2,980	Audrey (Betty) Elizabeth Cuppy	Elizabeth Young (1827)	Hannah Adelia Young	
2	44,113,851	53,913,635	11.8	3,300	Connie Edwards	Hendrick Schremling – Theobald Jung Sr.	Hannah Adelia Young	FTDNA
2	53,346,636	79,627,751	26.9	6,302	Jim Arnold	William McCormack (1836) & Mary Bain	Mary Bain	
2	56,360,803	78,713,382	23.5	3,040	Walt Gale	William Snadon (1700) & Catherine Snadon	Mary Bain	
2	81,873,176	101,977,153	8.6	2,336	Audrey (Betty) Elizabeth Cuppy	Elizabeth Young (1827)	Hannah Adelia Young	
2	82,363,176	106,011,094	11.1	3,257	Robert C Nelson	Elizabeth Young (1827)	Hannah Adelia Young	
2	82,456,901	119,943,392	22.1	5,845	Carolyn Vassos	Daniel Young (1755) & Eliz Windecker	Hannah Adelia Young	

2	82,919,371	106,016,513	10.8	3,250	Kay Montgomery Rumney	Elizabeth Young (1827)	Hannah Adelia Young	
2	116,142,763	127,910,398	11.1	1,361	Trisha MacLeod	Henry Young (1825) & Elizabeth Young	Hannah Adelia Young	
2	204,336,038	214,959,471	10.7	2,302	Jennifer Sewell Glover	Theobald Young (1691) & Maria C. Snyder	Hannah Adelia Young	
2	204,506,035	211,730,000	7.3	1,560	Kay Montgomery Rumney	Elizabeth Young (1827)	Hannah Adelia Young	
2	204,506,035	211,638,178	7.2	1,542	Dan Nelson	Elizabeth Young (1827)	Hannah Adelia Young	
2	204,507,942	211,784,597	7.3	1,554	Robert C Nelson	Elizabeth Young (1827)	Hannah Adelia Young	
2	204,524,820	211,362,876	6.9	1,475	Norm Sones	Elizabeth Young (1827)	Hannah Adelia Young	
2	227,720,427	233,653,809	9.8	1,430	Kay Montgomery Rumney	Elizabeth Young (1827)	Hannah Adelia Young	

<u>Genealogical Analysis</u>: Perhaps the most interesting group of segments yet discovered on the "p" arm of this chromosome begins about 33.5 Mb and ends about 44 Mb. This segment can be further attributed to an earlier generation, being inherited from Henry WINDECKER's mother Anna Elizabeth (WALRATH) WINDECKER (1716 Schoharie, New York – 1793 Ft. Plain, Minden Township, Montgomery County, New York). The origin of the segment can be narrowed down to the wife of Johann Georg WINDECKER, Anna Elizabeth WALRATH since while John Bowers is a descendant of both lines extending back from the daughter of their son Henry WINDECKER'S daughter; however Jody Hash Crowl descends only from the WALRATH family. Thus we can with some confidence attribute this segment to Anna Elizabeth's parents, Heinrich Conrad WALRATH born 1691 Runkel, Germany

died after 1746, when he wrote his will, in Canajoharie, New York (likely the Windecker Tract) and Anna Christina MATHEUS born 1685 Dunzweiler, Germany.

The segment between about 44 Mb to about 53 Mb has been inherited from Johann Adam JUNG 1717 Foxtown, Schoharie, New York – 1790 Seneca Township, Haldimand County, Ontario and / or his wife Catharine Elizabeth SCHREMLING (1720 Schoharie, New York – 1798 Barton Township, Wentworth County, Ontario). Connie Edwards and Maggie Cvek are descendants of both Hendrick and Maria Elizabeth (LANDGRAFF) SCHREMLING, and Johann Theobald (1691) and Maria Catharina (SNYDER) JUNG. The excellent genealogies help to identify the origin of the second of the two segments, 44 Mb to 53.9 Mb as YOUNG / SCHREMLING via Adam YOUNG and / or wife Catharine Elizabeth (SCHREMLING) YOUNG.

So in summary, the first segment can be linked to Elizabeth (WINDECKER) YOUNG; and the second segment originated with her husband Daniel YOUNG, the double great grandparents of Hannah Adelia (YOUNG) DAWSON.

Then there is a seamless transition (recombination point) that begins where the previous YOUNG – SCHREMLING segment left off at about 53 Mb and extends to about 80 Mb. It has been inherited by the author's Scottish BAIN ancestors via William SNADON born 1700 Alloa, Clackmananshire, Scotland and / or his wife and cousin Catherine SNADON born 1700 in the same place. The recombination must have occurred via the amalgamation of the anterior part of the chromosome of the author's great grandfather Joseph William DAWSON born 1872 Barton Township, Wentworth County, Ontario with the adjacent posterior part of the chromosome 2 of his wife Annie (MCCORMACK) DAWSON. With this sort of specific information it makes the genetic pedigree weave together with the genealogical pedigree. The chromosome at approximately 80 Mb shifts once again back to the YOUNG and WINDECKER family. Since all who share this segment with the author have both ancestral lineages it is not yet possible to parse it out to one or the other. The segment ends about 127 Mb, although it is possible that there is a small segment of about 10 Mb from another source (e.g., Mary BAIN) between 106 and 116 Mb. There is then a region whose origin is unknown until 204 Mb (near the end of the "q" arm of chromosome 2), extending to about 234 Mb which is also YOUNG via Johann Theobald JUNG (born 1691) and / or wife Maria Catharina SNYDER through their eldest son Johann Adam JUNG (born 1717).

It is important to note that the origin of each of these segments can be considered valid in that they have been "triangulated" with multiple matches to distant relatives (e.g., half 3rd cousins once removed; 7th cousins).

Summary and Conclusions: Considering the size of chromosome 2 it is quite amazing how much can be attributed to various early ancestors. To date there is no information to pinpoint the origin of the segment on which the HOXD genes reside. However the

mapping that is possible at this point shows some very clear junctions between the contributions of two of the great great grandmothers of the author – Hannah Adelia (YOUNG) DAWSON and Mary (BAIN) McCORMACK then back to Hannah Adelia (YOUNG) DAWSON – in the area between about 33 to 106 Mb. Secondarily, there is a similar close join is seen between the "contribution" of William and Mary (BAIN) McCORMACK and John DAWSON at about 7.5 Mb. Furthermore, at about 44 Mb there is a clear junction between the contribution of the Elizabeth (WINDECKER) YOUNG via her grandparents Hendrick Conrad WALRATH and Anna Christina MATHEUS; and a YOUNG segment (from Adam or Catharine Elizabeth (SCHREMLING) YOUNG), the parents of Daniel YOUNG extending to 53 Mb.

In all probability, over time, more of the maternal chromosome 2 of the author will be mapped; however to date all of the segments of known origin on the larger, q, end of the chromosome come from the author's great great grandmother Hannah Adelia (YOUNG) DAWSON. Hence the hypothesis to be tested is whether Hannah contributed the entire q end of chromosome 2 to the author – which will, alas, never be realized unless Ancestry.com offers a chromosome browser so that the large number of matches in their database will reveal the specific segments which are shared.

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