Understanding DNA in Genealogy



Peter Biggins, BY3164
Darien Library, Darien, CT
November 18, 2017

1953: Watson 25, Crick 37, Franklin 33



My BY3164 SNP

- 1953 paper published on the structure of DNA
- 1962 Nobel Prize awarded
- 2003 Human Genome Project completed
- 2008 My Y-DNA tested at FTDNA
- 2009 Clan Colla project started at FTDNA
- 2011 Clan Colla DNA presentation in Houston
- 2013 Z3000 SNP identified for Clan Colla
- 2015 BY3164 identified for Biggins & Bigham

Topics

- DNA 101
- Testing
- Three types of DNA
 - Mitochondrial mtDNA
 - Autosomal DNA
 - Y chromosome DNA
- What DNA can do for you

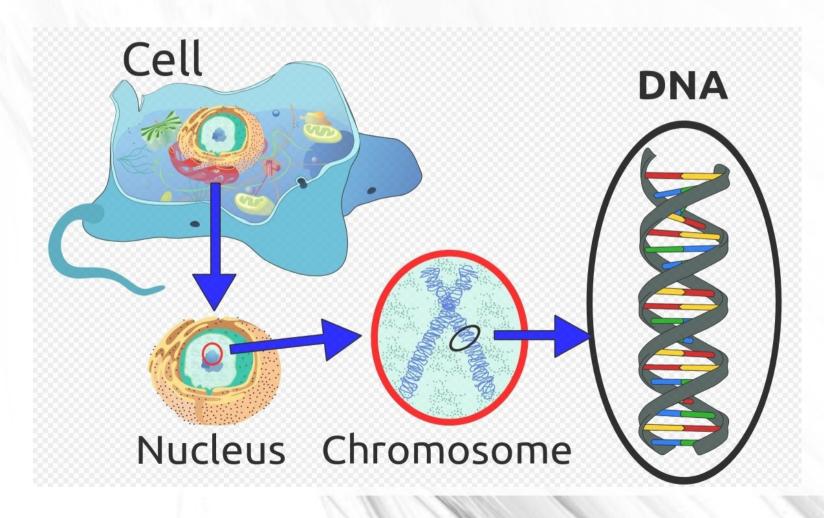
Housekeeping

- These slides are available at: mgs.darien.org/meetings.htm
- MGS has a DNA webpage at: mgs.darien.org/dna.htm
- Feel free to contact Peter at: pabiggin@optonline.net

DNA 101

- DNA: <u>Deoxyribo</u><u>Nucleic</u> <u>Acid</u>, a molecule found in the nucleus of cells
- STR: Short Tandem Repeats, a marker used to predict SNPs, e.g. 413b=24
- SNP: Single Nucleotide Polymorphism, a mutation on the Y chromosome, e.g., BY3164
- cM: centimorgan, a unit for measuring genetic linkage for autosomal DNA

DNA 101

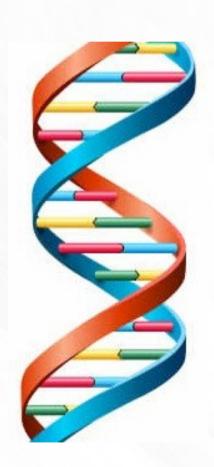


DNA 101

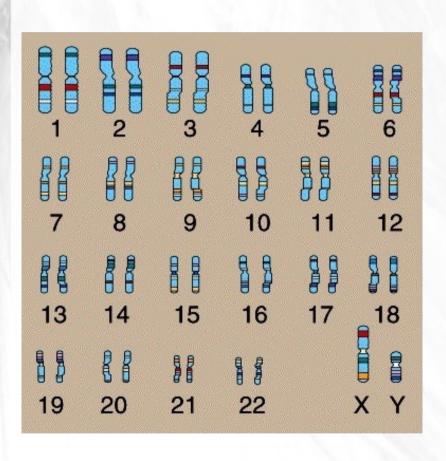
- You have 37 trillion human cells
- Almost every cell contains your whole genome
- The nucleus of each cell has 3 billion pairs of bases: A's, C's, G's, and T's
- These base pairs are spread among 46 chromosomes
 - Except, egg and sperm cells have only 23
 - X for female
 - Y for male

DNA 101: Base Pairs

- Bases of nucleic acids:
 - adenine (A)
 - thymine (T)
 - guanine (G)
 - cytosine (C)
- Base pairs:
 - A pairs with T
 - G pairs with C



DNA 101: Chromosome Pairs



- Pairs 1 to 22 are autosomes
- Pair 23 is the sex chromosome
 - XX for a female
 - XY for a male

DNA 101: Types of DNA

- In the cell's nucleus
 - Autosomal DNA is in the 44 non-sex chromosomes
 - Y Chromosome DNA is in the one male Y chromosome
- · In the cell's mitochondria
 - Mitochondrial DNA is found outside the cell's nucleus

DNA 101: Mutations

- Example (hg19): 8424676-C-A, aka BY3164
- On average, the genome for one person is 99.5% the same as another person
- Billions of cells die everyday
- Cells are constantly replicating by dividing
- · Variations occur, rarely, in replication
- These variations continue and are inherited
- DNA testing is based on these variations

DNA 101: Inheritance

- Mitochondrial DNA: 100% from mother
- Autosomal DNA: for each of pairs 1-22:
 - 50% chance from father
 - 50% chance from mother
- Y-DNA: 100% from father

DNA 101: Some Links

- The DNA double helix discovery
 - hhmi.com
- Human genome
 - Eupedia.com
- Genetic genealogy
 - ISOGG.org
- Y-DNA testing
 - lain's primer

Testing: 3 Types

Type	Testers	Ancestors
Mitochondrial DNA (mtDNA)	M and F	Mother to mother to mother, etc.
Autosomal DNA	M and F	All
Y chromosome DNA	M	Father to father to father, etc.

Testing: Sampling DNA

- Decide who is to be tested
- Decide the type of test
- Pick a testing organization
- Pick a specific test (and project)
- Order testing online
- Receive swabs by mail, swab, and return
- View results online

Testing: FTDNA Kit



Testing: Organizations

- Family Tree DNA autosomal, mitochondrial, Y chromosome
- Ancestry.com autosomal
- 23andMe autosomal, mitochondrial, Y chromosome
- Living DNA autosomal, mitochondrial, Y chromosome
- MyHeritage autosomal
- Yseq Y chromosome
- Full Genomes Y chromosome

Testing: Family Tree DNA

- Founded in 1999 by Bennett Greenspan in Houston
- All 3 types of DNA: autosomal, mtDNA, Y-DNA (STR and SNP)
- Saliva stored for upgrades and added types
- Projects administered by volunteers
- Public results on project webpage
- Annual conference for project administrators

mtDNA



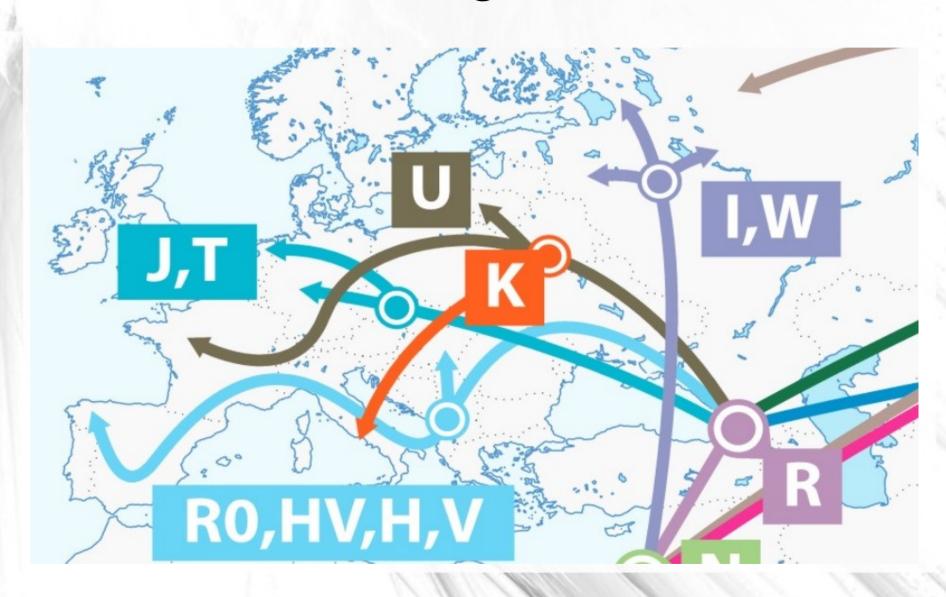
mtDNA

- The family tree of mothers
 - Both males and females have
 - Inherited from mother
- Can be used to verify two lines that go back mother-to-mother to the same woman
- Easiest to get from prehistoric remains
- Mutations occur much less frequently than Y-DNA

mtDNA: My results

- My oldest known maternal ancestor
 - My great great grandmother, Maria Anna Hauck (1792-1869)
 - From Königheim, 67 miles southeast of Frankfurt, in Baden-Württemberg
- I share U2e1a1c DNA with 10 people
 - 3 German out of 6,545
 - 1 Italian out of 4,916

mtDNA: Migration of U



mtDNA: Tests at FTDNA

- mtDNA Plus: \$99
 - 2 regions
- mtFull Sequence: \$199 (\$169 sale)
 - All regions

Autosomal

- Autosomes
- May be used by both men and women
- Two products
 - Matches mutations with other testers, measured in centiMorgans
 - Matches mutations with ancient populations around the world

Autosomal: Average cM

- Parent/child: 3600
- Sibling: 2650
- 1st cousin: 900
- 2nd cousin: 224
- 3rd cousin: 56
- Other distant cousins: 30

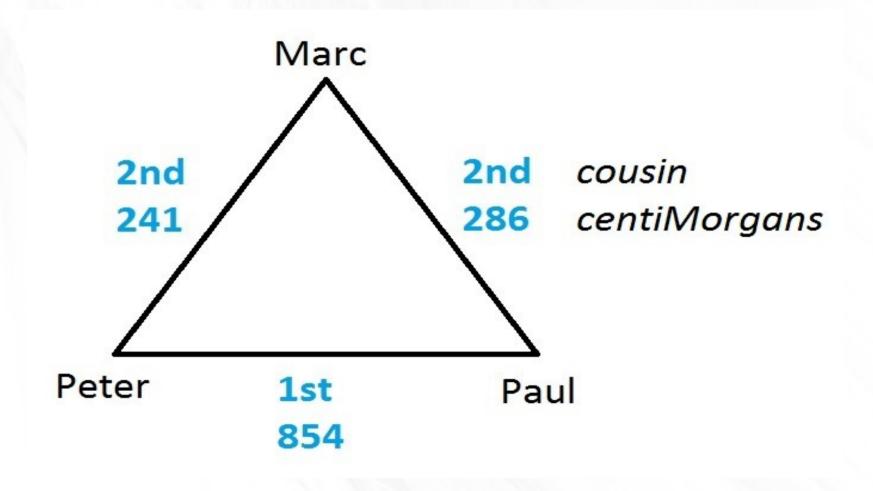
Source: The DNA Geek

Autosomal: My FTDNA matches

(CentiMorgans)

- 1st cousin Paul (854) already known
- 2nd cousin Marc (241) already known
- 2nd-4th cousin (108) unknown
- 3rd-5th cousin (35) with Beggan ancestor
- 5th-remote cousin (34), Beggan Y match
- 1,397 others (19-67) unknown

Autosomal: Peter, Paul, & Marc



Autosomal: My Origins

(50% Irish, 25% N. German, 25% S. German)

FTDNA

British Isles	57%
West and Central	26%
Europe	
East Europe	10%
Southeast Europe	4%
West Middle East	<2%
Central Asia	<2%

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MyHeritage

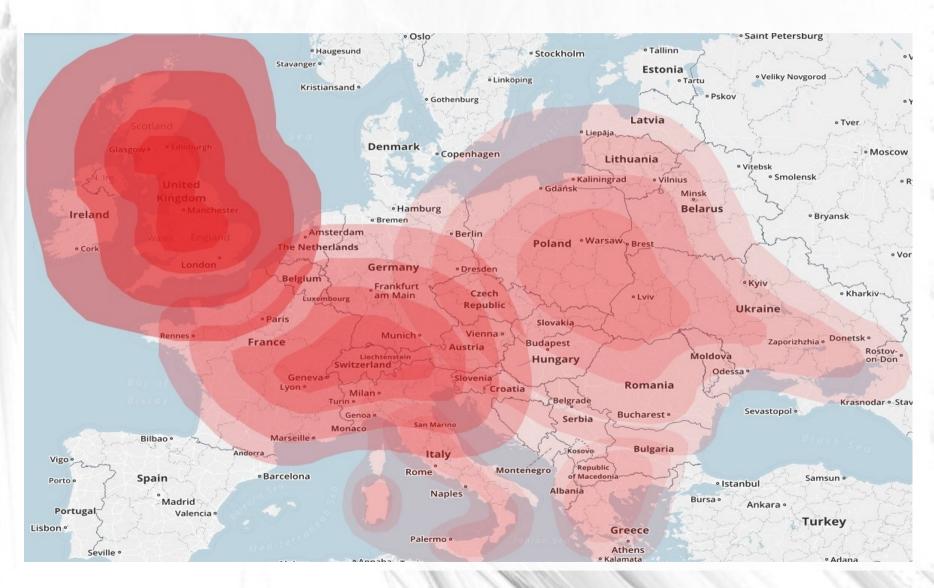
British Isles	69%
Scandinavian	12%
Finnish	1%
East Europe	16%
West Asia	<2%

Autosomal: 4 Swayne Sisters

Ancestry.com

	1	2	3	4
Ireland	45%	28%	33%	22%
Great Britain	16%	54%	42%	67%
Europe West	32%	5%	16%	<1%

Autosomal: myOrigins



Autosomal: My <2% West Middle East (Canaanite)

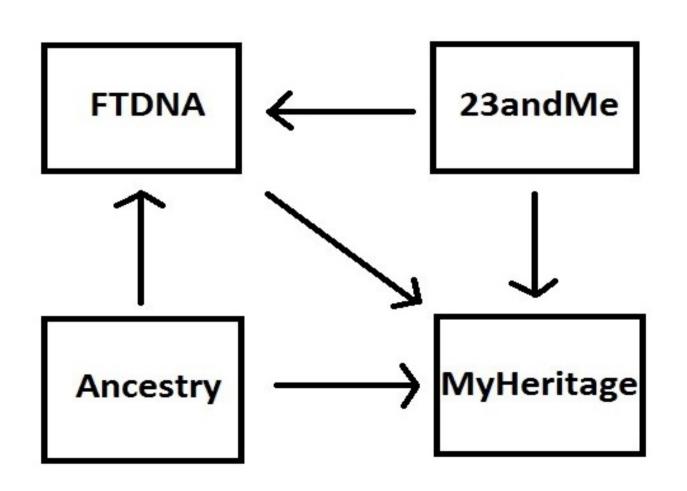


Autosomal: Recent Test Prices

- FTDNA: Family Finder \$59
- 23andMe: Ancestry \$69
- Ancestry.com: Ancestry DNA \$79
- MyHeritage: MyHeritageDNA \$59
- Living DNA: includes limited mtDNA and Y \$99

See ISOGG

Autosomal: Transfer options



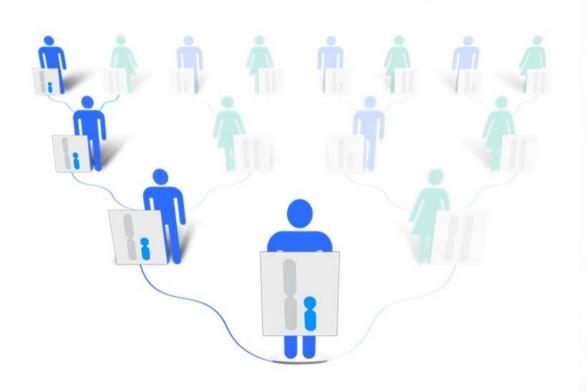
Autosomal: GedMatch

- GedMatch allows you upload DNA from Ancestry.com, FTDNA, WeGene, MyHeritage, 23andMe
- Also upload GEDCOM
- Variety of admixture models
- Multiple kit analysis

Autosomal: Says 'The Wirecutter'

- Ancestry.com is the best for:
 - ethnic roots
 - searching for contemporary relatives
- TV ads suggest a certainty that is well beyond the science upon which tests are based
- Ancestry.com is:
 - easier to understand
 - makes use of the largest database
 - is among the lowest-priced services

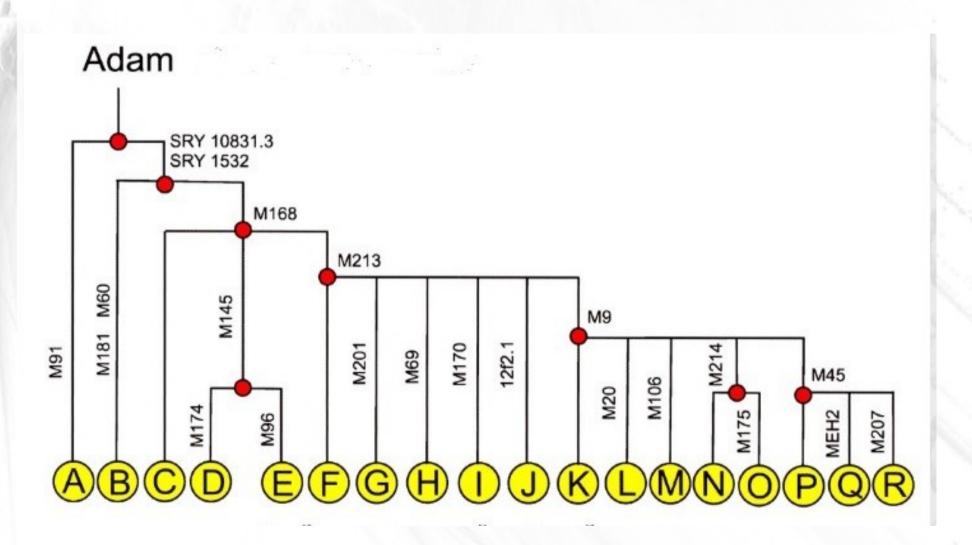
Y-DNA



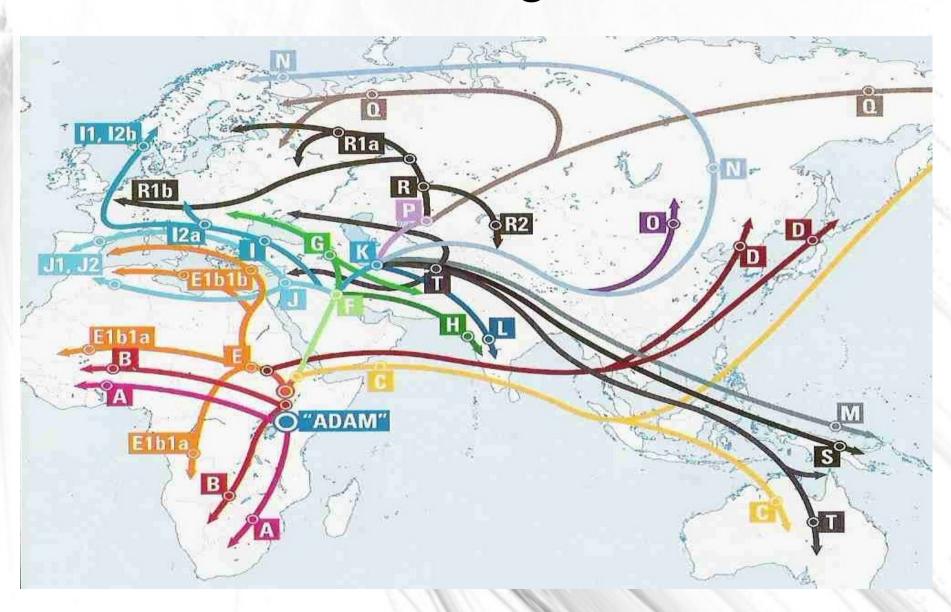
Y-DNA: for Men Only?

- Yes
 - Only men can be tested
 - Handed down from father to son
- But
- Many Y tests are bought by women
- They get Y-DNA from a brother, father, uncle, nephew, male cousin

Y-DNA: Haplogroups



Y-DNA: Migration



Y-DNA: About surnames

- Most surnames have variations in spelling (e.g., Cearbhaill, O'Carroll, Carrell, McCarroll, Carvill)
- Most surnames have a variety of DNAs (e.g., over 25 different Carroll DNAs)
- Surnames are not always inherited (e.g., adoption, fosterage, name change)
- Some surnames have a famous person (e.g., Charles Carroll of Carrollton)

Y-DNA: Solo Surnames

- Adoption
- Name change
- Small surname group, small sample
- Surname adopted long ago in a line that did not flourish

Y-DNA: Ancient Pedigrees

- Coincidence of surnames between ancient pedigrees and Y-DNA
 - The coincidence of names is not perfect
 - Ancient pedigrees don't pretend to capture all the names
 - They could easily have mistaken names
 - Many names are not very common, and only a small percentage of people have tested their DNA

Y-DNA: Project Types at FTDNA

- Surname: e.g., Biggins, Carroll
 - Multiple haplogroups
- Geographical: e.g., Irish, German
 - Multiple haplogroups
- Haplogroup: major SNPs, such as L21, DF21, Z3000

Y-DNA: STRs

- STRs: short tandem repeats, or mutations
- Tests: 12, 25, 37, 67, 111 STRs
- Unusual STRs can predict SNPs with varying degrees of confidence
- My STR tests
 - 2008: 67 STRs
 - 2011: 111 STRs

Y-DNA: Project Results Page

Kit Number	Name	Paternal Ancestor Name	Country	Haplogroup	DYS393	DYS390	DYS19	DYS391
I-M223, M284	4, L126 Isl							
	Byrne	William Byrne, b. 1864, Albany, New York	Ireland	I-L126	15	24	15	10
THE RESERVE OF THE PERSON NAMED IN	COLUMN THE PROPERTY OF THE	Type II/South Irish						
294201	Driscoll	John H. Driscoll b.1824 Kilbonane Co.Cork	Ireland	R-CTS4466	13	24	14	10
R-L21, DF13,	Z39589,	DF23, M222 Northwest Irish						
180395	Cavett	nelson cavett,b 1810,jackson,oh	Ireland	R-M269	13	25	14	10
252843	McDonnel	Daniel McDonald 1813 Ireland or Scotland	Ireland	R-FGC23592	13	25	14	11
R-L21, DF13,	Z39589,	DF41 S775, A600						
116493	Matthews	William Mathews, b.c. 1644, Possibly Wales or Holbu	England	R-A600	12	24	14	10
R-L21, DF21,	S971, Z3	3000, S953, BY3164 Clan Colla						
127469	Biggins	Patrick Biggins 1807 Co Cavan/Monaghan	Ireland	R-BY3164	13	24	15	11
146867	Biggins	James Biggins 1822 Co Monaghan	Ireland	R-M269	13	24	15	11
R-L21, FGC5	494, A114	48, A6342						
176456	Kenyon	John Kenyon, b. ~ 1600	United Kingdom	R-A1309	14	24	15	11
R-L21, L1065	Scottish	Cluster						
75783	Christie	Denis Christy b. c1722, Northern Ireland	Ireland	R-M269	13	24	14	10
R-P312, DF2	7, Y5058							33337/
198624	Carroll	Edmond Carroll, Stonepark, Limerick 1835-1906	Ireland	R-Y5061	13	25	14	11

Y-DNA: Match Page

67 MARKERS - 68 - MATCHES					
Genetic Distance ↑	Name	Earliest Known Ancestor	Y-DNA Haplogroup		
2	Mr. Adrian James Beggan		R-M269		
3	Keith J. Bigham A T Y-DNA67		R-M269		
5	Mr. Joseph Willis Plunket Property Plunket Mr. Joseph Willis Plunket	Joseph Plunket b1828 LA d 1910-20	R-M269		
5	Mr. Jarvis Elbert Plunkett	Joseph Plunkett b1828 LA d 1910-20	R-M269		
5	Mr. Gerard Beggan ⚠	Beggan	R-L21		
5	Mr. Mark Wayne Bigham TO Y-DNA111	Hugh Bigham 1750-1831 Ireland to Lebanon, PA	R-BY3164		

Y-DNA: STR Matches

Variation in Matches Among MGS Testers at 67 STR Markers

Don	30
Len	43
Marc	3
Peter	68
Pete	21
John	182

Y-DNA: My STR Matches

GD is genetic distance

- 853 GDs of 0-1 at 12 STRs
- 554 GDs of 0-2 at 25 STRs
- 48 GDs of 1-4 at 37 STRs
- 68 GDs of 2-7 at 67 STRs
- 6 GDs of 7-10 at 111 STRs

Y-DNA: My 111 STR test

- Z3000 Colla SNP predicted by unique STRs
 - 47th STR 511=9
 - 48th STR 425=0
 - 76th STR 505=9
 - 91st STR 441=12
- BY3164 Biggins SNP predicted by unique STR
 - 50th STR 413b=24

The Fermanagh Story by Peadar Livingstone, 1969

BEGGAN (Ó Beagáin): also Little. This family was common in the Clones-Roslea-Donagh area. It was originally Ó Beacain and this was often anglicised Little. Little was itself a British name and some of the minor planters bore it. O'Donovan mentions Beggan under Clones and there are many Beggan inscriptions in Donagh cemetery. There are 139 Little voters in Fermanagh today, and 44 Beggans or Beagans.

Y-DNA: STR 413b=24

2006	Beaghen	1860 Ireland to Brooklyn
2006	Little	Co. Cavan to Scotland to Australia
2007	Bigham	Colonial Pennsylvania
2008	Biggins	1835 to Canada, then Illinois
2009	Biggins	1840 Monaghan to Illinois
2010	Bigham	Colonial Pennsylvania
2010	Beggan	Clones, Co. Fermanagh
2011	Beggan	Clones, Co. Fermanagh
2016	Biggin	1849 Somerset, England, to Australia
2017	Biggins	1830 Co. Monaghan to Scotland

Y-DNA: SNPs

- SNP: single nucleotide polymorphism, or mutation
- Tests: individual SNP, SNP pack, Big Y
- SNPs can be predicted by STRs
- SNPs, inherited like surnames, define the family tree of your paternal line
- My SNP tests
 - 2011: DF21
 - 2014: Big Y

Y-DNA: Big Y

- "Full genome" testing by Family Tree DNA
- Covers a large portion of the Y chromosome
- Branches defined by two or more men having the same SNP
- Trees constructed by volunteers, FTDNA, YFull
- Private SNPs will form new branches if shared with future testers

Y-DNA: BY3164 SNP

- My "terminal" SNP the one I share with the fewest number of men
- At 8424676 on the human genome, a C changed to an A (hg19)
- One of 5 SNPs shared with Mark Bigham
- Occurred around 1600 AD
- Others named Beggan, Beaghen, Little are predicted to have it based on STR 413b=24

Y-DNA: Six with BY3164



Y-DNA: Unique Big Y SNPs, hg19

Bi	qc	ins	127	7469
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8424676-C-A

9646350-G-C

14358670-A-C

14839640-G-A

17705431-C-T

18019516-G-GTA

18105747-C-CT

18647271-C-A

Bigham N86783

8424676-C-A

9646350-G-C

14358670-A-C

14839640-G-A

18647271-C-A

19454555-TA-T

21983582-G-A

22521329-G-T

23072046-T-C

Y-DNA: Shared Big Y SNPs

Biggins 127469

8424676-C-A

9646350-G-C

14358670-A-C

14839640-G-A

17705431-C-T

18019516-G-GTA

18105747-C-CT

18647271-C-A

Bigham N86783

8424676-C-A

9646350-G-C

14358670-A-C

14839640-G-A

18647271-C-A

19454555-TA-T

21983582-G-A

22521329-G-T

23072046-T-C

Y-DNA: SNP Tree

Biggins/Bigham

8424676-C-A

9646350-G-C

14358670-A-C

14839640-G-A

18647271-C-A

Biggins 127469

17705431-C-T 18019516-G-GTA 18105747-C-CT

Bigham N86783

19454555-TA-T 21983582-G-A 22521329-G-T 23072046-T-C

Y-DNA: Named SNPs

Biggins/Bigham

BY3164

9646350-G-C

BY3167

BY3169

BY3181

Biggins 127469

17705431-C-T 18019516-G-GTA 18105747-C-CT <u>Bigham N86783</u>

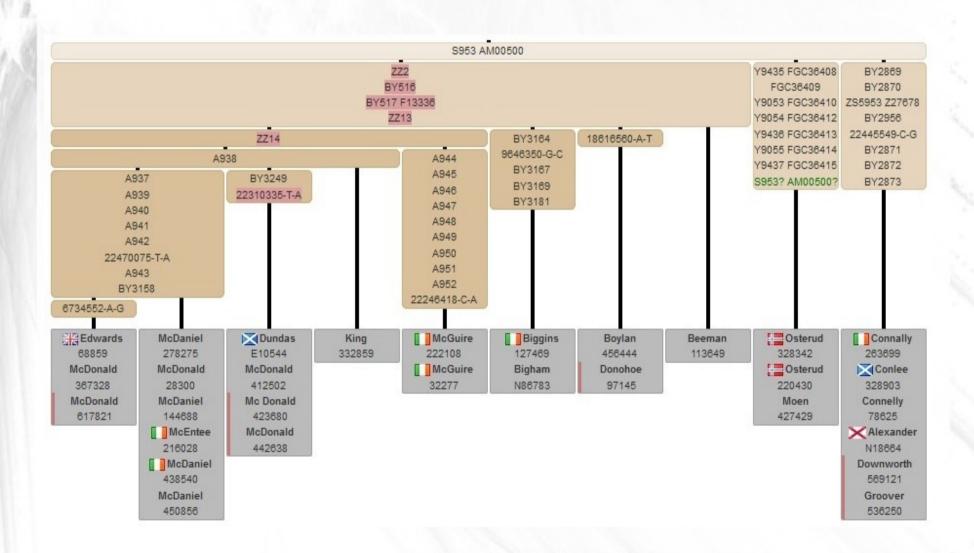
19454555-TA-T

21983582-G-A

22521329-G-T

23072046-T-C

Y-DNA: S953 Tree



Y-DNA: Dating & Placing SNPs

- A SNP has no recorded date or place of birth
- SNPs occur randomly and infrequently
- E.g.: 14 men have the A938 SNP. They have
 4 to 16 downstream SNPs. Average 9.3
- $9.3 \times 120 = 1116 (900 AD)$
- Archeological evidence helps
 - Test DNA from bones found in a specific place
 - Carbon-date bones

Y-DNA: Rathlin 1 Man

- In 2006, scientists at Queens University Belfast and Trinity College Dublin uncovered the bones of a man in a Bronze Age cist
- To the rear of McCuaig's Bar on Rathlin Island
 - 25-minute ferry ride from the Glens of Antrim
 - 11 miles from the Mull of Kintyre in Scotland
- Carbon-dated back to 2025-1885 BC
- Y-DNA testing found the DF21 SNP
- Indicates the date of DF21 around 2700 BC

Y-DNA: Bones at McCuaig's Bar





Y-DNA: My Deep Ancestry

Ancestral SNP Group	# SNPs	Gener- ations	Time (rough)	Place	Men 2016
Z3000	20	80	80 AD	Isles	80
Z3006	1	4	200 AD	Isles	60
Z3004	2	8	440 AD	Ulster	56
S953	1	4	560 AD	Ulster	23
BY516	4	16	1040 AD	Ulster	14
BY3164	5	20	1640 AD	Ulster	2
17705431-C-T	3	18	1820 AD	Ulster or US	1

Y-DNA: The Three Collas

- Lived in Ulster in the 4th century A.D.
- Remarkable coincidence between two sets of surnames:
 - Men with Z3000 DNA
 - Men mentioned in ancient histories of Collas
- Carroll, Connolly, Devine, Hart, Higgins, Hughes, Kelly, MacDougall, McAuley, McClain, McDonald, McGuire, McKenna, McMahon, McQuillan, Monaghan

Y-DNA: The Three Collas

- 2 McMahon 4th cousins have traced their ancestry back to Colla da Crioch
- 4 McDonald 5th cousins have traced their ancestry back to Colla Uais

Y-DNA: Historical Pedigrees

- R-S781: Sir John Stewart of Bonkyl
- R-S8350: House of Wettin
- R-L746: High Stewards of Scotland
- R-YP326: John of Islay, Lord of the Isles

Y-DNA: Historical Pedigrees

- R-Z30233: Rathlin 1
- R-BY20010: Charles Carroll of Carrollton
- R-Z3000: Clan Colla
 - A77: McMahon, Lords of Dartry
 - A938: Somerled, MacDonnell of Antrim and Leinster

Y-DNA: Historical Pedigrees

- R-M222: Niall of the Nine Hostages
- R-Y5058: Breassal Breac
- R-FGC5659: Brian Boru
- R-L1403: Seven Septs of Laois
- R-CTS4466: Eoghanachta, Corca Laidhe
- R-FGC6545: Hy Maine

Y-DNA: History Debunked

- Clan Colla was not related to
 - Niall of the Nine Hostages
 - Hy Maine
- Ely Carroll, Brain Boru, and Eoghanachta were not related to each other
- Lords of the Isles were not descended from Somerled

Y-DNA: My Testers

- Peter Biggins
- Sean Biggins our great great grandfathers had adjoining farms in Illinois
- Paul Drueke maternal 1st cousin
- Marc Matthews maternal 2nd cousin
- Michael Patrick Carroll wife's 2nd cousin
- Michael McDonnel wife's 3rd cousin

Y-DNA: My FTDNA Projects

- 2008: Biggins surname started by Daniela Maneta (N=22)
- 2009: Z3000 Clan Colla (572)
- 2011: Z16291 Ely Carroll (100)
- 2012: Drueke, CTS10893 Saxon (25)
- 2012: Carroll surname preexisting (366)
- 2014: Middlesex Genealogical Society (18)
- 2015: Y5058 Breassal Breac (102)

Y-DNA: MGS Project at FTDNA

P312, L21, DF13 (4000-3000 BC)							
	Z39589			FGC5494	FGC11134		
DF49 Z2980 Z2976 DF23 Z2961 M222 Niall (100 AD)	L1335 L1065 Little Scottish Cluster (300 AD)	DF41 S775 A600	S971 Z3000 Z3004 Clan Colla S953 BY516 BY3164	FGC5561 A1149 A1148 A1308 A6342 A1309	A353 Z16250 CTS4466 Irish Type II (100 AD)		
		21149398	17705431	14515146			
1810 Ohio	1722 Ireland	1640 UK	1807 Cavan	1600 UK	1824 Cork		
Don Cavett	Len Christie	Marc Matthews	Peter Biggins	Pete Kenyon	John Driscoll		
		Biç	g Y (present tim	ne)			

Y-DNA: Community

- FTDNA projects
 - Members
 - Administrators
- Yahoo groups
- Anthrogenica
- Citizen scientists
- Big Tree (P312)

Y-DNA: STR Tests at FTDNA

- Short Tandem Repeats
- Predict genetic distance from other testers
- 5 STR tests (sale price)
 - Y-12: \$39
 - Y-25: \$124
 - Y-37: \$169 (\$129)
 - Y-67: \$268 (\$229)
 - Y-111: \$359 (\$299)

Y-DNA: SNP Tests at FTDNA

- SNP: Single Nucleotide Polymorphism
- Specifies one's place on the Y-DNA family tree
- Tests (sale price)
 - Individual SNPs: \$39
 - SNP packs: \$119 (\$101)
 - Big Y: \$575 (\$475 + free upgrade to Y-111)
 - Confirms predicted SNPs
 - SNPs down to the present time
 - Present and future matches

Y-DNA: Suggested Approach

- Test men in different lines of your family
- Join surname projects at FTDNA. If none, start one and become an administrator
- Buy Y-67 (STR) and Big Y (SNP) on sale
- Contact your matches
- Join haplogroup (SNP) projects
- Join Yahoo groups, and submit Big Y raw data
- Look for ancient pedigrees, surname variants

What DNA can do for you

- Confirm genealogical research
- Break down a brick wall
- Find distant cousins
- Suggest ethnic make-up
- Explain a surname
- Create a genetic pedigree
- Connect with an historic genealogy