Using DNA Testing to Explore NBGS Spring Seminar Series - April 29, 2017 Brian Cassidy - bpc@unb.ca Www.cassidys.ca

#### CBC News – September 27, 2016

## Genetically modified baby born with DNA from 2 women

Mother is a 36-year-old woman who had 4 pregnancy losses and 2 deceased children

CBC News Posted: Sep 27, 2016 2:53 PM ET Last Updated: Sep 27, 2016 4:17 PM ET



A sperm is inserted directly into an egg during an IVF procedure. In this case, nuclear DNA from the donor's egg was removed and the mother's added, before fertilization. (CBC)

- 'It scares people off': Why Canada isn't ready to talk about '3-parent' babies
- Britain votes to allow world's first '3-parent' babies

DNA is a hot topic in today's genealogy magazines



New England Historic Genealogical Society<sup>™</sup>

#### Objectives and Agenda

• The objective of this presentation is to explain my family's experience with DNA testing and to try and remove some of the mystery around what can be a dauntingly complex science

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- This presentation should help you decide if DNA testing is right for you
- Agenda
  - DNA for Genealogists The Theory
  - Break
  - Our DNA Testing Experience at Family Tree DNA (FTDNA)
  - Conclusions and Recommendations

– Questions?

#### What Is DNA?

- deoxyribonucleic acid
- Double helix formed by base pairs attached to a sugar phosphate backbone
- DNA is the component of human cells which contains genetic information passed from generation to generation
- This genetic code is very similar for closely related people
- Every cell in our body has DNA except red blood cells
- We receive DNA from both our mother and father

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#### Chromosomes

- We all have 23 pairs of chromosomes within a cell nucleus
  - The 23<sup>rd</sup> chromosome is the sex chromosome
  - Of each pair, one is inherited from the mother and one from the father
  - The mother's contribution is always X
  - The father's contribution is X or Y
    - If X, then XX becomes a girl
      - Girls do not have a Y chromosome
    - If Y, then XY becomes a boy
      - Boys have both X and Y chromosomes

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#### Y-Chromosome DNA (Y-DNA) and Markers

- Only men have a Y chromosome
  - Passed from father to son

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- Y-Chromosone DNA provides information about the direct male line
- Markers are testing locations on the Y chromosome
  - Markers have technical names like DYS#391 and DYS#439



Thymine (Yellow) = TGuanine (Green) = GAdenine (Blue) = ACytosine (Red) = C

#### Y-Chromosome Marker Mutations

- Mutations are small changes in the DNA sequence
- Mutations take place at random intervals

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- Typical timeframe is once in about 500 generations (10,000 15,000 years)
- Mutations are helpful in identifying family tree branches
  - This indicates the ancient origin of one's major population group (haplogroup)

#### Mitochondrial DNA (mtDNA)

- This DNA is outside the nucleus of the cell
- We (both males and females) inherit mitochondrial DNA from our mother and *none from our father*
- mtDNA mutations (like Y-DNA mutations) occur very slowly over thousands of years
- mtDNA mutations allow us to identify unique population groups (haplogroups) along the direct maternal line
- Both men and women can do mtDNA testing



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#### Human Origin and Migration

- Our human species (Homo sapiens) evolved about 200,000 years ago in central Africa during a time of dramatic climate change
- From this point of origin, we have migrated around the world
- Migrations within Africa are the oldest (130,000 to 200,000 years ago) and migrations to the Pacific Islands and New Zealand are the most recent (1,000 to 10,000 years ago)
- Migrations to Europe
  - Eastern Europe 30,000 to 55,000 years ago

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- Western Europe 10, 000 to 30,000 years ago
- Scotland and Ireland 1,000 to 10,000 years ago
- Major population groups, called *haplogroups*, can be identified by studying the DNA mutations of people living in these areas

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• This same science can be used to predict migration paths

### Haplogroups

- *Haplogroups* are major population groups
- Can be established along male (Y-DNA) or female (mtDNA) lines
- Listed alphabetically (A, B, C etc.)

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- Major groups include Europe, Africa, Asia, Pacific Islands, the Americas
- Subgroups of haplogroups are called *subclades* and this can further refine one's ancestral origin

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• A *macro-haplogroup* is a collection (or super group) of closely associated haplogroups

#### Haplogroups (mtDNA Migration Map)



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#### Haplogroups (Y-DNA Migration Map)



 $\text{tree: } \textbf{Y-DNA Adam} \rightarrow \textbf{A} \ \textbf{B} \ \textbf{DE} \ \textbf{C} \ \textbf{F} \qquad \textbf{F} \rightarrow \textbf{G} \ \textbf{H} \ \textbf{IJ} \ \textbf{K} \qquad \textbf{K} \rightarrow \textbf{LT} \ \textbf{NO} \ \textbf{MS} \ \textbf{P}(\rightarrow \textbf{Q} \ \textbf{R})$ 

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# SNPs and their role in determining genetic disorders

• SNP – single nucleotide polymorphism

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- SNPs are a common genetic variation (anomaly / mutation) among people
- They represent a difference in a single DNA building block
- SNPs normally occur between genes but when located near or in a gene, they may play a direct role in disease by affecting the gene's function.
- SNPs can predict a person's reaction to certain drugs and risk of certain diseases
- There are roughly 10 million SNPs in the human genome
- SNPs in conjunction with one's haplogroup information can help refine one's ancestral origin

#### Autosomal DNA Testing

- This is testing of the first 22 (non-sex) chromosomes
- Each of these chromosomes has a different number of genes
  - For example #1-2800 and #22-750
- We inherit autosomal DNA from both parents, 4 grandparents, 8 great-grandparents etc.
- An autosomal DNA test can confirm relationships with a high degree of accuracy for parent/child and all relationships up to the second cousin level.
- For all relationships other than parent/child, additional contextual and genealogical information is required to confirm the nature of the relationship.
- For relationships beyond second cousin, DNA samples are required from many family members starting with the oldest family members available, then aunts, uncles and cousins etc.

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• See also http://isogg.org/wiki/Autosomal\_DNA

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- This article includes a YouTube video on autosomal DNA testing

#### Enough Theory Already!

• Time for a break after which we will look at a practical DNA testing example using Y-DNA and autosomal DNA testing



#### Our Brick Wall

Are David and Peter Cassidy related?

Trying since 1995 to connect these two family trees Can DNA testing help?



#### **DNA** Test Objectives

- Learn about DNA as it relates to genealogy research
- Try to establish a solid connection between Peter Cassidy and David Cassidy
- Prepare a presentation for others (which forces one to learn and understand)

#### Y-DNA Testing and two more definitions...

- Y-DNA testing is the most common form of genealogical DNA testing
  - This is the search for the MRCA (most recent common ancestor) in the direct male line
  - This test can predict your (paternal) haplogroup
- It's key to success is the close correlation between surnames and Y chromosomes
- *Male only testing* since only men have a Y chromosome
- As stated previously, markers are the testing points along the Y chromosome
- At each marker, a short DNA code repeats itself

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- This short section of code is called an *STR* (*short tandem repeat*)
- The number of times an STR repeats is called the *allele value*

#### Finding a Match

- The probability that two people are related is determined by comparing the allele values at each marker
- This matching service is provided by the DNA testing organization (in our case, FTDNA)
- The result is a comparison chart (called a Y-DNA TiP Report) showing generation by generation the probability that two people are related
- A *genetic distance* is assigned

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- Genetic distance is the number of differences or mutations between two sets of Ychromosome DNA or mitochondrial DNA test results
- The lower the genetic distance, the more likely two people are related
- A person has a genetic distance of zero with their siblings (identical test results)

#### Y-DNA Testing at Family Tree DNA

- Peter Cassidy set up an account at familytreedna.com and ordered a Y-DNA37 kit (37 markers) on July 18, 2015
  - Kit consists of two cotton "toothed" swab kits
    - <u>https://www.youtube.com/watch?v=AN07Adpu7Bs</u> (You Tube How to Test)
- Requested personal information included the name and location of the oldest known male ancestor (Andrew Cassidy 1762-1846 County Donegal, Ireland)
- Kit number 430909 was assigned by Family Tree DNA
- Results obtained September 18, 2015
  - The basic results are a Y-DNA Certificate showing the markers and allele values

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#### Certificate – Y-DNA

This Certificate confirms that you have had your DNA analyzed by Family Tree DNA. The outcome from each of the thirtyseven Loci examined is reported in the table below.

For your benefit we have listed the Locus designation for all thirty-seven Loci utilized by the geneticists supporting our company. If your alleles for the thirty-seven Loci match another person exactly, then you share the same Haplotype.

Family Tree DNA is a genealogical tool designed to aid individuals wanting to "connect" to other relatives lost in time and where the paper trail no longer exists.

#### **Peter Cassidy**

Your Kit #	430909										
Allele	DYS393	DYS390	DYS19	DYS391	DYS385	DYS426	DYS388	DYS439	DYS389-1	DYS392	DYS389-II
	19	23	10	10	13-19	() <b>H</b> ()	13	10	13	12	30
Allele	DYS458	DYS459	DYS455	DYS454	DYS447	DYS437	DYS448	DYS449		DYS464	
	16	8-10	11	11	26	16	20	30	12-15-15-15		5
	DYS460	GATA-H4	YCAII	DYS456	DYS607	DYS576	DYS570	CDY	DYS442	DYS438	
Allele	10	11	19-21	16	14	18	17	34-39	13	10	

### Y-DNA TiP Report Summary

The probability that these people share a common ancestor with Peter Cassidy within a certain number of generations

David Cas Genetic Di	sidy istance 1	Mark T. Ca Genetic Di	issidy istance 3	William L. Genetic Di	Cassidy stance 3	Alan Cassidy Genetic Distance 3		
Generations	Percentage	Generations	Percentage	Generations	Percentage	Generations	Percentage	
2	59.37	2	29.55	2	29.74	2	36.26	
4	83.49	4	59.06	4	59.32	4	74.10	
6	93.29	6	78.28	6	78.50	6	89.48	
8	97.28	8	89.08	8	89.23	8	95.73	
10	98.89	10	94.70	10	94.79	10	98.26	

- Our current research goes back only 5 generations.
- We have no explanation for the fact that Alan Cassidy has a genetic distance of 3 but almost the same MRCA percentages as David Cassidy.

#### The Family Trees with TiP Report Percentages

Given the 27 year difference in birthdates for George and Andrew, the MRCA is likely in generation 7 (95.73% probability).



#### Observations from Peter's Y-DNA37 Test

- Our (paternal) haplogroup is I-M223 (FTDNA) or I2a2a (ISOGG)
  - Europe / Ireland

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- Peter and David Cassidy are **almost certainly related** within 6 or 7 generations
- It is worthwhile for both David and Peter to **continue digging** through ancestral records in an attempt to establish a hard connection
- There are currently 5 close matches with Peter's DNA in the FTDNA database but David Cassidy is the most promising with a genetic distance of 1

#### Observations from Peter's Y-DNA37 Test

- Still no hard link between Peter and David, so David's tree information cannot be added to our Cassidy Family database
- Now that Peter's DNA is registered with FTDNA, we expect to find other matches in the future as people submit their DNA for testing
- FTDNA sends Peter an email when a new match is discovered

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– Two new matches in the past two years but none with genetic distance 1

#### FTDNA Website – Results and Information

- Y-DNA Testing Results
  - Print Y-DNA Certificate
  - View and/or download Y-STR results
  - View close DNA matches with other persons in the FTDNA database
    - Relations along the paternal line in the search for the MRCA
  - View haplogroup information
  - View ancestor migration map

#### FTDNA Website – Results and Information

- Family Finder Results (autosomal DNA test)
  - View close DNA matches with other persons in the FTDNA database
    - First and second cousins etc.
  - Chromosome Browser (tool for comparing DNA segments with others)
  - Family Matching (sorting confirmed matches along maternal and paternal lines)
  - myOrigins (mapping one's ethnic and geographic origin)
  - ancientOrigins (ancient ancestral migration routes)
- Other Resources
  - Forums (various topics including in-depth DNA research)
  - Family Projects (Cassidy Family Project)
- <u>FTDNA</u> website (www.familytreedna.com)

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#### Cassidy DNA Project

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- FTDNA partnership with World Families Website
- Various surname projects
- People share DNA information and discuss results in a forum
- The Y-DNA results page is interesting but we have not yet figured out how to incorporate this into our research

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- Our kit (430909) is labelled **Haplogroup I Possible Lineage I**
- The Cassidy DNA Project forum is not very active or useful
- <u>Cassidy DNA Project (www.worldfamilies.net/surnames/cassidy)</u>

#### Are We Related?

• Our family website: <u>www.cassidys.ca</u>

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- Operating since 2001 with our own home grown Linux operating system and public domain genealogy software
- Hardware based for many years at Peter's home near Nashville, TN
- In 2013, server hardware outsourced to Amazon Web Services (cloud web service)
- The Genealogy Section has a **Research Guide**
- The Research Guide has an **Are We Related** section which helps people determine if their family may be related to ours
- The **Are We Related** section has been expanded to include information on DNA Matching (our experience and some general information on the subject)
- View the Are We Related page at our family <u>website</u> (www.cassidys.ca)

#### Reasons why one might want to do DNA testing

- Determine if two (male) individuals are closely related (usually same surname)
  - Y-DNA testing
  - This is classic genealogical DNA testing
- Determine one's ancient origins along paternal lines (haplogroup)
  Y-DNA testing
- Determine one's ancient origins along maternal lines (haplogroup)
  - mtDNA testing
- Connect with living relatives and validate uncertain relationships
  - Autosomal DNA testing
- Determine if one has any inherited medical conditions; genetic risk factors; responses to certain medications and traits such as lactose intolerance?

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- Autosomal DNA - SNP testing

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#### DNA Testing Risks You may get surprising or disturbing results

- You may discover that your ancient heritage is not what you have been told it is
  - <u>http://on.wsj.com/2dc2jM0</u> (Wall Street Journal Report)
- You may discover that you are not related to people with whom you assumed were related
- For autosomal SNP medical tests, are you psychologically prepared for the news that you may be at high risk of some very serious disease?

#### **DNA** Testing Websites

• Y-DNA and mtDNA and autosomal

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- ftdna.com (Family Tree DNA)
- dna.ancestry.ca
- Autosomal SNP (Medical)
  - 23andme.com
  - decode.com

#### How much does a DNA test cost?

- These prices from Family Tree DNA (ftdna.com)
- Y-DNA (men only)
  - 37 markers US\$169
  - 67 markers US\$268
  - 111 markers US\$359
- mtDNA
  - Basic US\$79
  - Full Sequence US\$199
- Autosomal
  - Family Finder US\$79

#### **Conclusions and Recommendations**

- Begin your DNA testing with another individual with whom you are trying to establish a relationship
- Research your family tree first!
  - The better one's family tree is known, the better the chance of establishing a link to other family members who are also doing DNA testing
  - This exercise caused us to carefully re-examine our oldest genealogical records and we did find errors and make changes
  - Our best source for old records in Ireland was baptismal records dating back to 1805
  - Reviewing old information can reveal new treasures

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- The Y-DNA37 test (37 markers) is sufficient to establish paternal (surname) relationships
- Autosomal DNA testing is a great way to validate close family relationships

#### **Conclusions and Recommendations**

- We have found genealogical DNA testing to be fun, educational, interesting and useful
- To date, we have made no additions to our family tree but we have an ever growing number of promising leads which encourages us to continue with grass roots research to establish solid connections
- DNA testing may not break down your brick walls but it should put a crack in them
- DNA testing is yet another modern day tool in your genealogy toolkit
- Some knowledge of this subject is compulsory for any 21<sup>st</sup> century professional genealogist

#### Sources, References and Credits

- Research Information
  - Family Tree DNA
    - DNA Testing
    - Understanding DNA
    - Understanding your Y-DNA37 Results
  - New England Historic Genealogical Society
    - American Ancestors Newsletters (various articles on DNA)
      - PANB Containers 26672 and 69193
  - International Society of Genetic Genealogy (ISOGG) (isogg.org)
  - CBC News
  - Wikipedia Genetic Genealogy
- Photographs and Charts
  - Family Tree DNA
  - Getty Images
  - Wikipedia
- Credits
  - New Brunswick Provincial Archives Staff
  - Peter Cassidy and David Cassidy (DNA contributors)

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#### Questions?



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