DNA Basics

Using DNA for Family History

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Glossary

- (see http://edmund-rice.org/dnagloss.htm)
- DNA = Deoxyribonucleic Acid = blueprint of life
- Base = Nucleotide = smallest unit of DNA, one of four possible units, denoted as A, C, G, or T
- Marker = Locus = any identifiable feature on the DNA chain
- STR = Microsatellite = marker consisting of repeating sequences, characterized by the number of repeats, e.g., 15

Glossary (cont.)

- Mutation = any change in DNA sequence
- Chromosome = major grouping of DNA, visible at certain stages of cell division
- Y Chromosome = determines maleness
- mtDNA = Mitochondrial DNA = DNA found in cell components responsible for energy production (maintained outside the cell nucleus) – *much* slower mutations than Y STRs

Why Y? Why mtDNA?

- Usable for genealogy
  - Inherited from just one parent, no confusion
  - Inherited from just one grandparent, etc.
  - Traces an entire lineage
- Contrast with the rest of the DNA
  - Comes in pairs, one copy from each parent
  - The pairs are reshuffled and randomly subdivided at each generation -- may have no genes at all from a given distant ancestor

Rice DNA Projects

- (see http://edmund-rice.org/haplotype.htm)
- 549 members tested as of Sept. 17
  - 471 tested at FTDNA for Y DNA
  - 57 tested elsewhere for Y DNA
  - 8 tested at FTDNA for mtDNA
  - 13 tested elsewhere for mtDNA

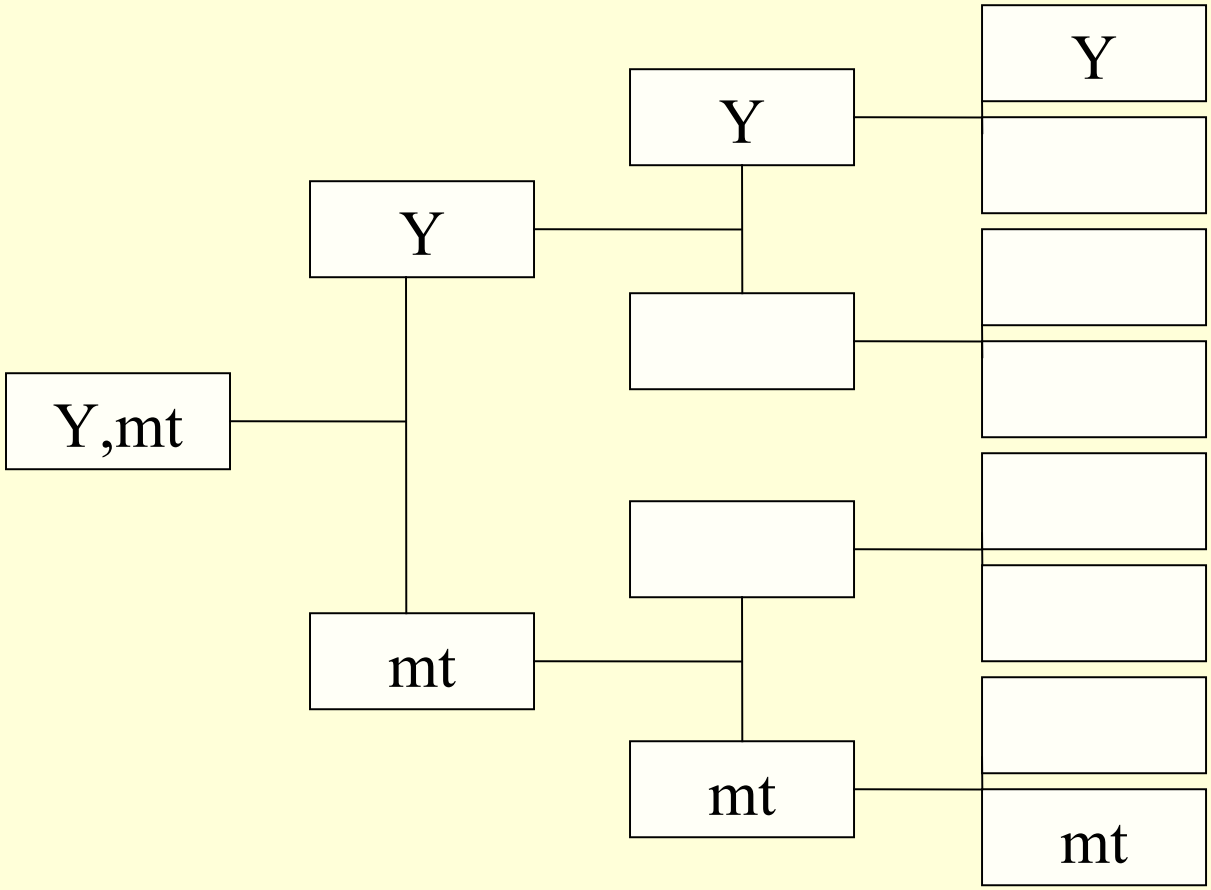
DNA Applications

- Exciting parts
  - Genealogy
  - “Deep Ancestry” (beware!)
- “Scary parts” (not relevant here)
  - Forensics (criminal id or paternity)
  - Health screening (diagnosis or prediction)

Genealogical DNA

- Male lines
  - Y DNA parallels surnames
  - Rice DNA project, for example
  - Crucial need also for conventional genealogy
- Female lines
  - mtDNA inherited only from the mother
  - Crucial need also for conventional genealogy
- Mixed lines
  - Limited applicability

DNA on the pedigree



Y DNA Genealogy

The Rice project is one of thousands of active surname projects at FTDNA. As a matter of fact, there are 3 different projects for SMITH, plus 1 for SCHMIDT. There are also projects for Allen, Bates, Chandler, Drury, Ewing, Farmer, Grant, Hastings, and so on (sample of names of ERA members). Generally, any male with the specified surname, or a variant, is eligible for membership in a project.

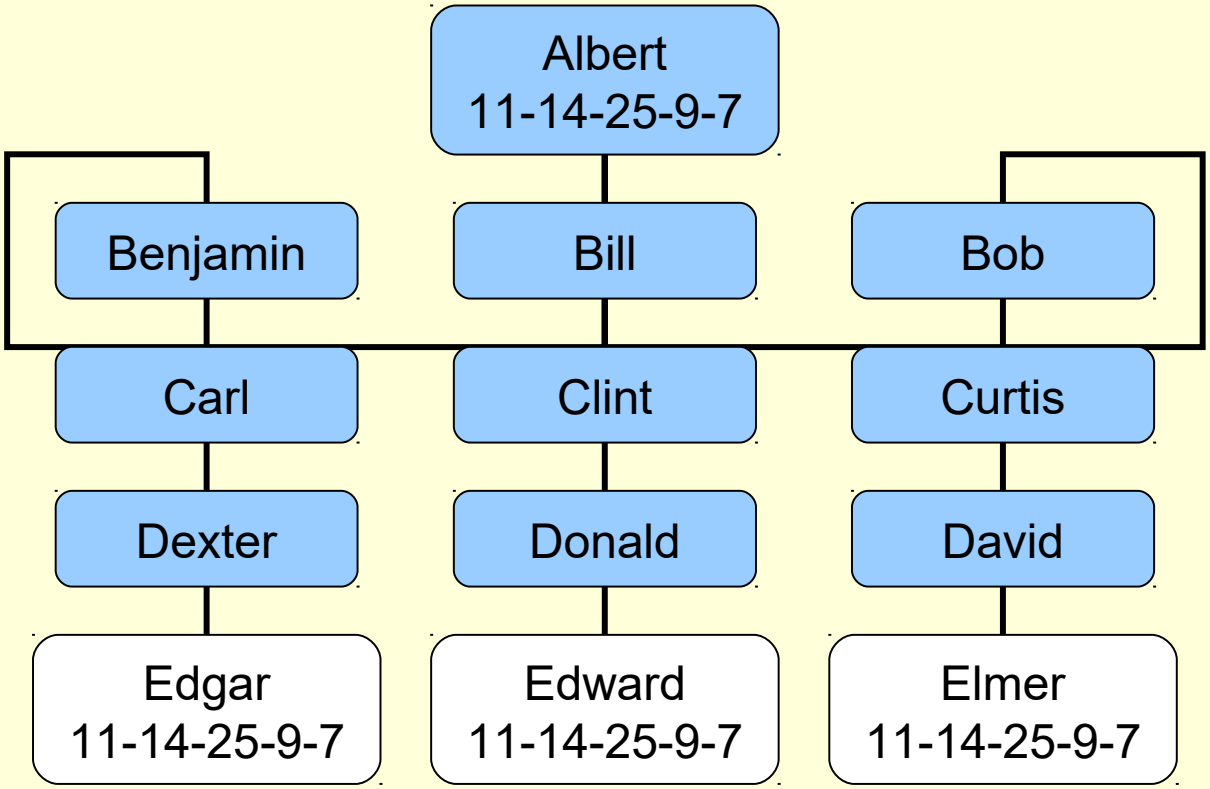
Y DNA Examples

1. Research validation – support or refute connections based on limited evidence
2. Lineage organization – discover at last which families are related
3. Desperately seeking cousins – breaking through “brick walls”
4. Wide-open exploration – all of the above

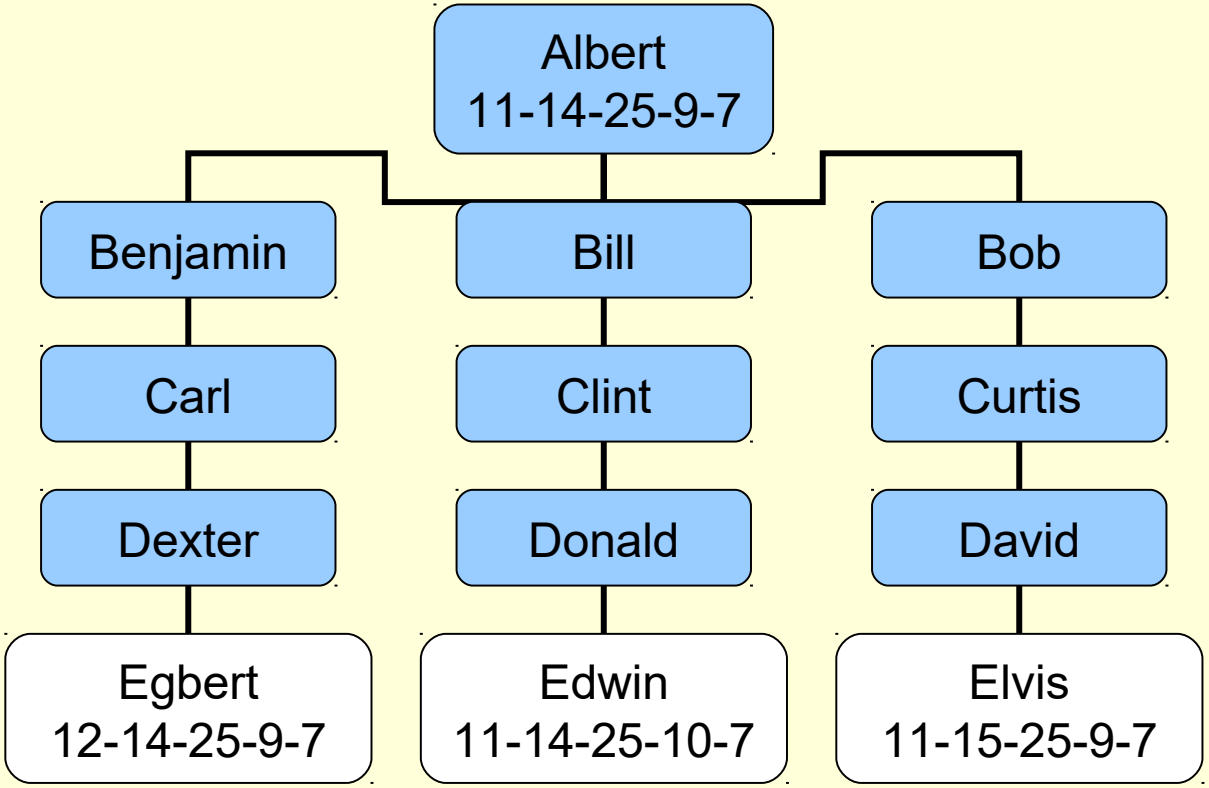
Y DNA Tools I

- Comparison of DNA patterns among testees who are “known” to be related.
- This applies to (1) Research validation.
- Assume, as in the Rice project, that we start with volunteers who have documented lineages back to a common ancestor.
- We examine the numeric results, looking for agreement.
- Need more conventional genealogy if mismatch.

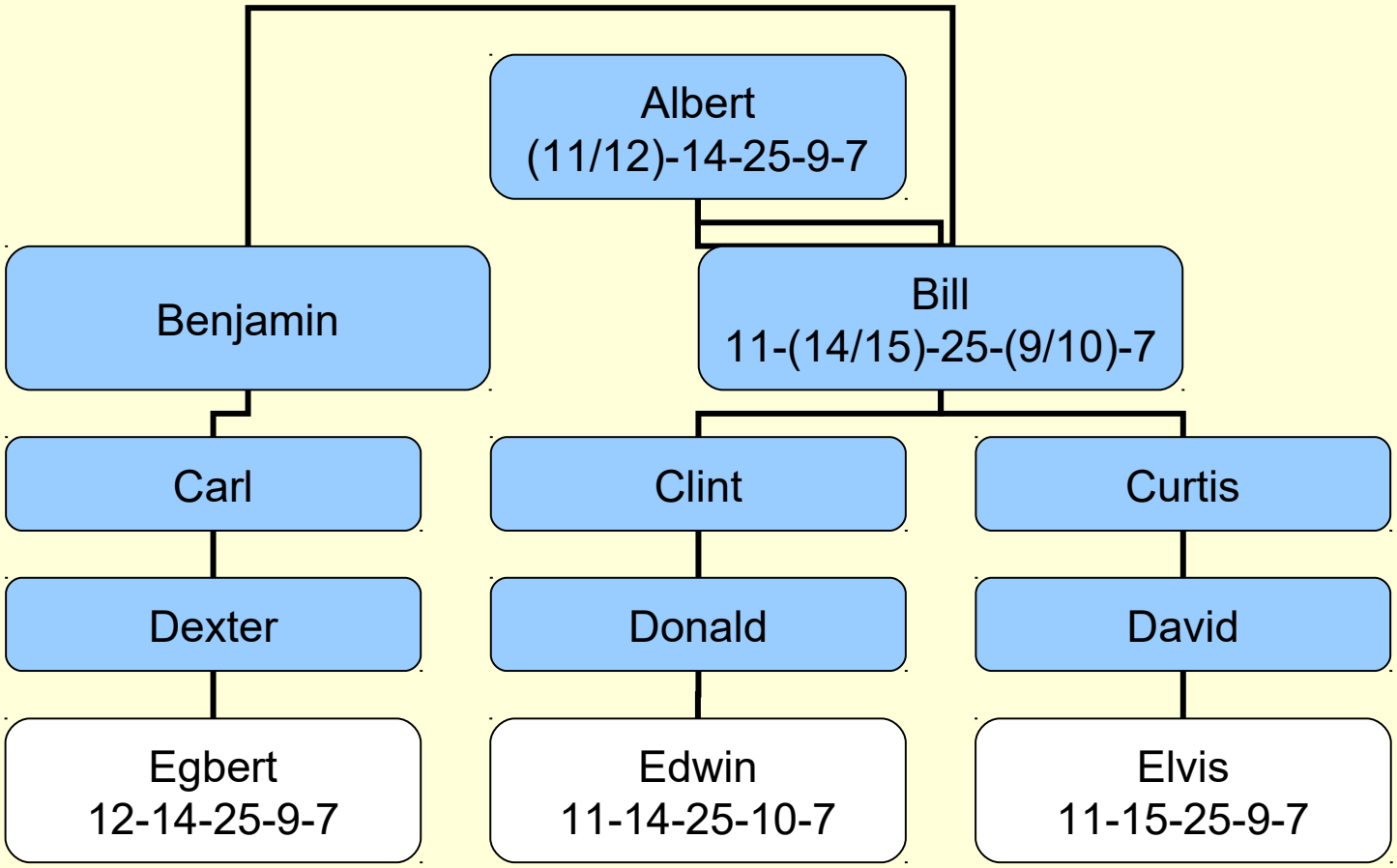
Reconstructed Ancestral Patterns (unanimity)



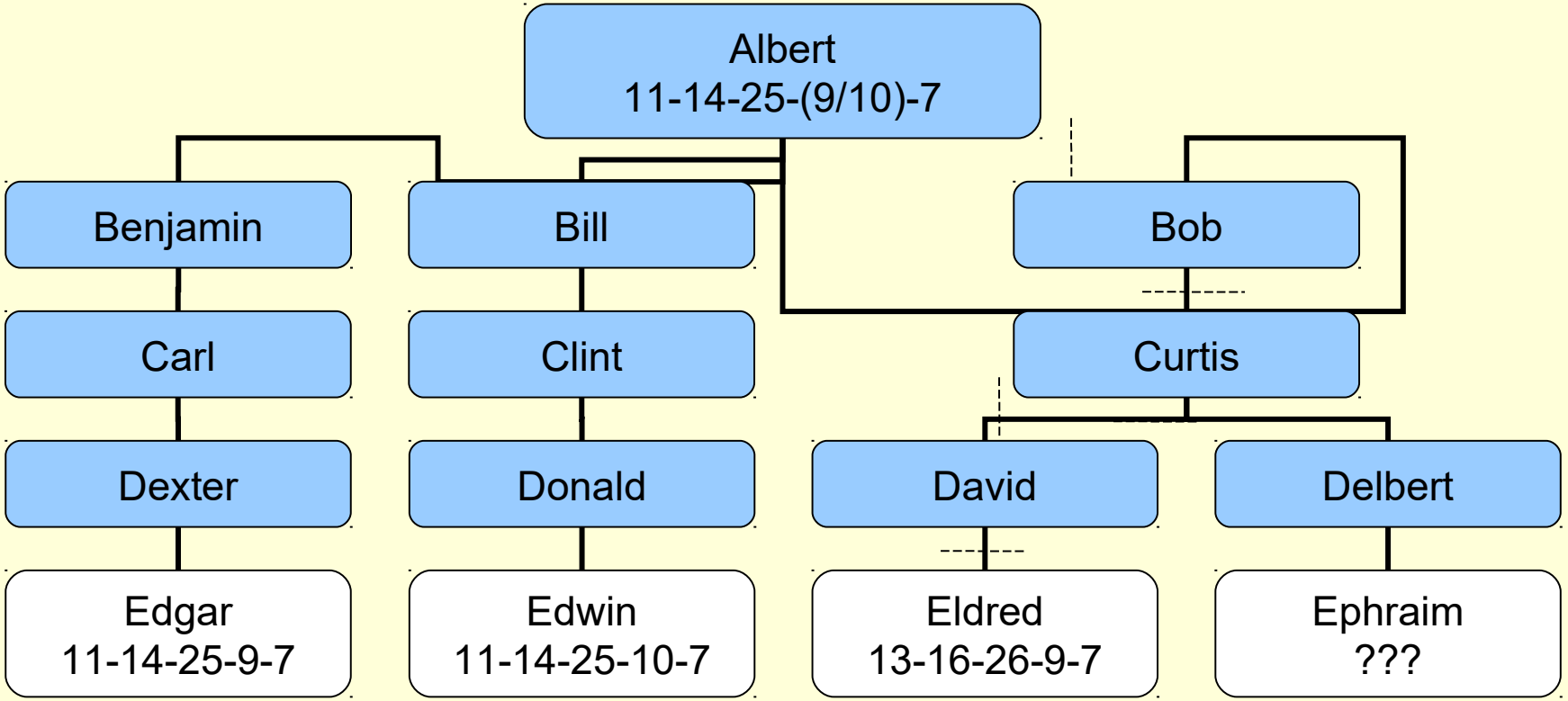
Ancestral Patterns II (majority rule)



Ancestral Patterns III



Ancestral Patterns IV (surprise!)



Summary of dummy DNA results

Edgar	11	14	25	9	7
Edward	11	14	25	9	7
Edwin	11	14	25	10	7
Egbert	12	14	25	9	7
Eldred	13	16	26	9	7
Elmer	11	14	25	9	7
Elvis	11	15	25	9	7
Ezekiel	11	14	25	10	7

Y DNA Tools II

- Comparison of DNA patterns between a known tree and a new testee.
- This applies to (2) Lineage organization, and (3) Desperately seeking cousins.
- Assume that we have a reconstructed ancestral pattern and now test Ezekiel as 11-14-25-10-7.
- We *may* be able to assign him to a branch.

Y DNA Tools III

- If you are female, recruit a male relative to take the DNA test on your behalf
- This can apply to any of the DNA examples

mtDNA Genealogy

- Everyone has mtDNA, not just females, but males do not pass mtDNA to children
- Essential to have conventional research done in advance (or be very lucky)
  - no natural gathering places for probable kin
  - lower resolution in mtDNA test results
  - maternal line research is harder

mtDNA Genealogy (cont.)

- Test results expressed as differences from a standard mtDNA sequence
- Each mtDNA base is numbered (1-16569)
- HVRI = ~16001-16569, HVRII = ~1-574
- Substitution – 16519C
- Deletion – 524-
- Insertion – 315.1C

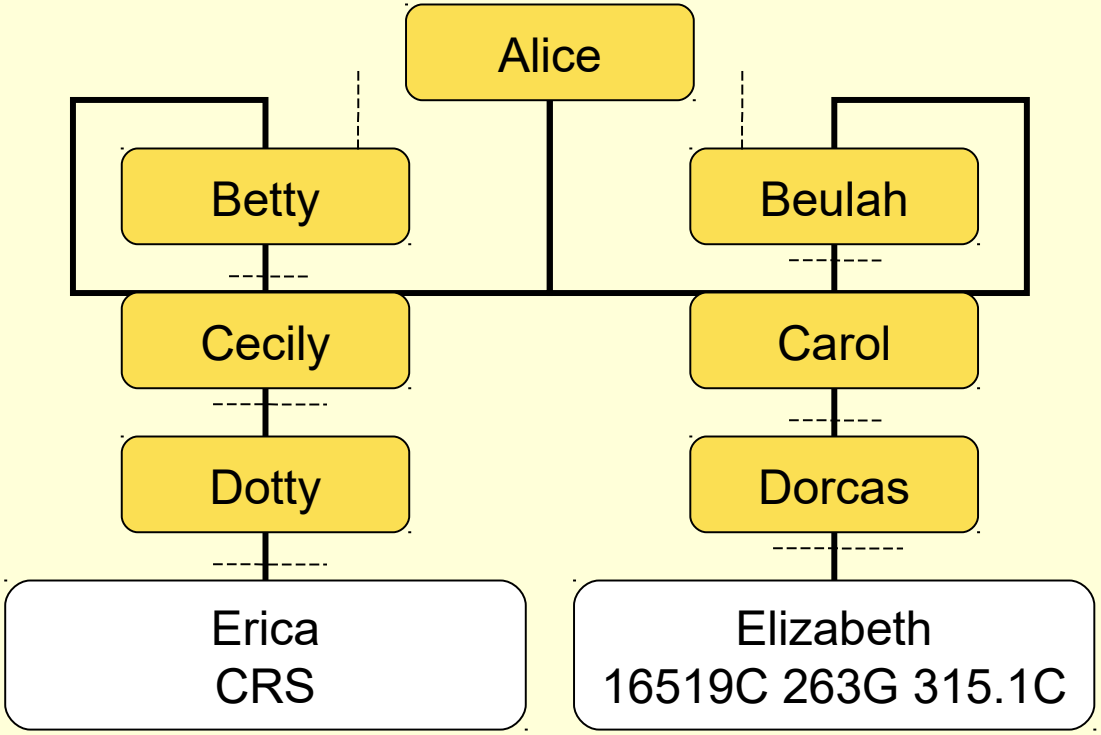
mtDNA Examples (almost same)

1. Research validation – support or refute connections based on limited evidence
2. Lineage organization – **NOT**
3. Desperately seeking cousins – people with recent “brick walls”
4. Wide-open exploration – all of the above

mtDNA Tools I

- Comparison of DNA patterns among testees who are “known” to be related.
- This applies to (1) Research validation.
- Look for another female-line descendant of your own ancestor.
- Compare test results. They should match.
- Possible outcomes similar to those for Y DNA.

Ancestral mtDNA pattern? (surprise!)



mtDNA Tools II

- Comparing a new testee to a known tree
- Not yet realistic – too few trees known
- We are still mostly at “Tools I” stage