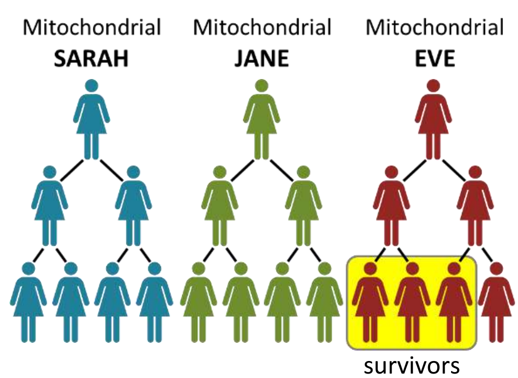
MITOCHONDRIAL EVE

(1) The majority of people believe that half of their DNA comes from their mother and half from their father, but this is not entirely true. Of the genomic DNA (the DNA found inside the nucleus), 23 chromosomes come from your mother and 23 come from your father. This is an equal contribution, but when you look outside of the nucleus, there is more DNA found in the cytoplasm. Mitochondria, that supply your cells with energy, also have DNA. Each mitochondrion contains a small loop of DNA that it uses to self-replicate. All the mitochondria in your cells come from your mother, so mitochondrial DNA (mtDNA) is all maternal, hence just a little more than 50% of the DNA in your cells come from your mother.

(2) How did all of your cells get populated by only your mother's mitochondria? This distribution of mitochondria occurred during fertilization. An egg has a huge cytoplasm that contains the numerous organelles needed to kick-start cell division, and this includes many mitochondria. In contrast, the sperm is very small and has very few mitochondria which are used merely to power the tail. The average egg contains 10 000 000 copies of mtDNA while a sperm cell contains only 100 copies of mtDNA. This greatly decreases the chances of any paternal mtDNA contribution. Also, after fertilization, only the sperm's chromosomes and centriole remain because enzymes in the egg destroy the rest of the sperm, including all of its mitochondria. This leaves the zygote (fertilized egg) with mitochondria that come solely from the mother's egg.

(3) The discovery of mitochondrial DNA was made in the 1960s and in the decades that followed, more research was done that led to some interesting findings. In 1987, a group of geneticists, led by Rebecca Cann, published a research paper in the Journal *Nature*, which concluded that all humans on Earth today can trace their ancestry back to a common female ancestor. She lived in Sub-Saharan Africa (likely around Tanzania) 100 000 to 200 000 years ago. During this time, *Homo sapiens sapiens* (modern anatomical humans) were developing as a distinct species different from the other species of humans, like *Homo neanderthalensis,* that existed at the same time.

(4) This most recent common female ancestor of all modern humans was named "Mitochondrial Eve". She was not the only female human alive at the time, but only Mitochondrial Eve and her descendants contributed to the gene pool of modern humans. It is proposed that at some point in the past the human population experienced a genetic bottleneck. This means that some catastrophic event decimated the human population and reduced it to a small number of surviving individuals who were all descendants of Mitochondrial Eve, and who all had her



mitochondria. The findings of Rebecca Cann's research team also supports the *Out of Africa Theory* which claims that modern humans originated in Africa and through waves of successive migrations, came to populate different areas of the globe.

(5) What evidence supports this? In Rebecca Cann's study, she recruited 147 people across all of the major racial groups. Examination of their mtDNA allowed researchers to trace back every racial group to Mitochondrial Eve. This is

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made possible because mitochondria replicate asexually. Unlike genomic DNA which is recombined in every generation through sexual reproduction (i.e. every offspring is genetically different from their parents), mtDNA is merely cloned when new mitochondria is made. Thus everyone in your family has different genomic DNA, but you, all of your siblings and your mother will share the same exact mtDNA (though your father's mtDNA is not represented in his offspring).

(6) Mitochondrial DNA only changes if a genetic mutation occurs in the mtDNA of the egg cells resulting in offspring. Thus mutations are rare and only show up once in every 1000 generations. Hence, you most likely have the same mtDNA as your mother's mother 1000 generations back. However, when mutations do occur, the result is that every generation going forward will contain mitochondria with changed mtDNA. These changes give clues to how different human populations have evolved over time. If population A and population B have similar mtDNA, this means that few mutations have occurred between them and that not long ago, the two separate populations were most likely one population. However, if population A and population B have many more mutations between their mtDNA, this indicates they diverged from one another further in the past. Comparing differences in the mtDNA of populations allows researchers to trace backwards to the origins of the first humans. Not surprisingly, within themselves, African populations have the greatest differences in their mtDNA because they have the oldest genetic lineage.

Article Questions

1) What two factors make it unlikely that paternal mitochondrial DNA will be found in the zygote?

2) In the diagram in paragraph 4, why is each human generation represented only by female symbols and not by both female and male symbols?

3) Who was Mitochondrial Eve?

4) What is the Out of Africa Theory?

5) How did studying the mtDNA of various races help trace them all back to Mitochondrial Eve?

6) Though there are differences in mtDNA amongst modern humans, modern chimpanzees show much more diversity (many more differences) in their mtDNA. What does this suggest about chimpanzee evolution?

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