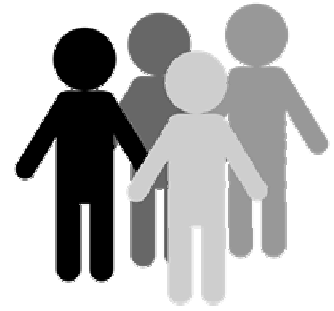


# Maryland Essays in Human Biodiversity



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# Maryland Essays in Human Biodiversity

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# Editors' Corner

## Smallpox: From eradication to warfare

*Soroush Rais-Bahrami*

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Smallpox is a very important virus in the news today but it has a long history that has been too easily forgotten. Because of its long history as a human pathogen worldwide, extensive research and manipulation led smallpox from an epidemiological tragedy to an eradicated evil; but now there is a prevalent fear that a widespread biological attack could cause an outbreak for which the world is unprepared.

Smallpox has been infecting people for hundreds of years. No one is positive as to when it first emerged, but some scientists believe it originated in Arabia around 572 AD, but its name was coined in France centuries later where it was seen as a milder version of syphilis (Bazin 2000). Syphilis became known as the great pox and subsequently the name smallpox was born (Hopkins 1983). Due to the simple transfer of this disease, many people throughout history have suffered the effects of this deadly virus. Some of the most notable victims are the Egyptian Pharaoh Ramses V and Queen Elizabeth I of England (Stearn and Stearn 1945). Smallpox does not select against the rich or the poor, the famous or the unknown. It has been responsible for the elimination of many languages and has been a significant factor in the conquests of many cultures. Without the breakouts of smallpox, Cortez might not have conquered the Aztecs, and Pizarro might have been unable to overcome the mighty Inca Empire. Besides infecting the people of new and unexplored regions, smallpox also spread like wildfire across Europe. During the Middle Ages, a high percentage of the population was infected, and when Europeans came to the Americas to colonize, smallpox annihilated the Native American population. The Native Americans had never been exposed to any disease quite like smallpox and their immune systems were unable to overcome the invasion leading to a near extermination.

With the smallpox epidemic raging across Europe, millions of people died or suffered horrible disfiguring scars if they survived the infection. This continued until Edward Jenner developed a vaccine to combat the smallpox virus in the latter 18th century (Tortora *et al.* 2001). Jenner's development of this invaluable vaccine stemmed from the serendipitous discovery that milkmaids did not acquire smallpox. He linked this immunity to their continual exposure to cowpox, a closely related virus that infects bovine hosts. To test his theory on the relatedness of cowpox

and smallpox, Jenner took a small scraping from a cowpox blister on the hand of Sarah Nelmes and injected the pus into the skin of James Phipps, an 8-year-old boy. Phipps developed a few raised bumps at the site of inoculation, but never developed a full outbreak of cowpox or smallpox. Following this experiment and a few more related studies, Jenner attempted to publish his finding through the Royal Medical Society. Disbelieving of such a new and radical idea, the Royal Medical Society rejected Jenner's paper and he was forced to publish privately (Flint *et al.* 2000).

The eradication of the smallpox virus involved a long battle fought by many individuals, in many countries, most of whom received no credit for their efforts. Even though the World Health Organization (WHO) was in charge of the effort to eradicate smallpox, they were not originally supportive of the idea. In 1958, when the WHO held their annual meeting in Minneapolis, Minnesota, the proposal to eradicate smallpox was suggested by a Russian named Victor Zhdanov. The WHO made minimal attempts to see this mass vaccination carried out, but did not put much effort toward the goal until 1965 when the major nations of the world established resolution demanding the elimination of smallpox in a ten-year span. This resolution demonstrated to the WHO that the eradication of smallpox was the highest priority of the major powers around the globe. (Shurkin 1979)

In an effort to gain more support from the United States and Russia, the WHO appointed D.A. Henderson to lead the effort. Henderson had previously been the head of the Center for Disease Control's (CDC) Epidemic Intelligence Service (Shurkin 1979). While in charge of the largest, most skilled group of epidemiologists in the nation, Henderson was appalled at the CDC's inability and inexperience addressing smallpox as a public health issue. Thus, he began dispatching teams to every outbreak in Europe to observe and learn all they could (Henderson 1999; Shurkin 1979). From the CDC, Henderson moved on to lead the effort to deliver mass measles vaccinations to people living in African countries, which brought him to the attention of the WHO and made him the perfect candidate for heading up the worldwide effort to eradicate smallpox (Shurkin 1979). Without Henderson's dedication and tenacious actions, smallpox would never have been eliminated.

When the smallpox vaccine was first developed, it required serial infection of human subjects in order to be maintained, which eventually led to its ban due to the spread of syphilis and hepatitis associated with the serial infections. Since the strain of smallpox could no longer be used, it was replaced with the vaccinia virus, whose origin is unknown (Flint *et al.* 2000). Currently, there is no treatment if a person becomes infected; however the vaccine is fairly effective in protecting people from infection. The only problem with the vaccine is that it requires boosters every few years or the individual's immunity will diminish over time.

Currently smallpox is effectively non-existent in the public, global arena. No cases have been reported in over twenty years, and since 1985 smallpox vaccinations have not been used for the general public (Bazin 2000). Vaccinations in the United States stopped between 1971 and 1972, and thus anyone currently under the age of thirty is susceptible to a possible smallpox infection (Bazin 2000). The youngest generation of people around the world has no immunity to the virus since they have not received the vaccine and have not lived through the viral infection. Subsequently, this could lead to a potential future epidemic.

In order for the smallpox virus to reemerge into the population it would require human assistance. To the best knowledge of the scientific community there are no remaining animal reservoirs in which smallpox could be hiding, so the only possible way for this disease to surface is for smallpox to take over a new niche once it has been introduced into the environment again. The only real danger of smallpox resurfacing comes from any leftover samples kept in research labs or undisclosed stocks elsewhere. At one point there were over one hundred laboratories with live smallpox samples for the purpose of research, but most have been destroyed in order to prevent the virus from falling into the wrong hands (Winslow 1974). Presently only two known small vials of the smallpox virus remain for research. One is housed at the CDC in Atlanta, Georgia and the other is stored at the Institute of Virus Preparations Moscow Russia; both locked up in liquid nitrogen freezers at minus 94°C. Because of the potential damaging consequences should smallpox ever emerge again, the WHO has 500,000 doses of the Variola vaccine on hand in event of an outbreak with the capability of producing more in a matter of weeks (Stephenson 2001). There are no other known stocks of the virulent smallpox virus or agents that can cause an epidemic, but there are threats of using smallpox as a biological weapon of terror.

There are documented cases where smallpox has sprouted in isolated environments such as the case of Janet Parker. She died on September 11, 1978, in London, from a smallpox infection acquired from a strain that was kept in a lab in the same building where she worked (Shurkin 1979). Her infection demonstrates that vaccinations do not last throughout a lifetime and booster shots are necessary for continued immunity. This confirms the notion that most of the world's current population is now susceptible to smallpox because many people have not been vaccinated,

and those who have been vaccinated have not received regular booster shots.

Since smallpox has been effectively eradicated and poses such an epidemiological threat to the general population that has no immunity to the virus, it is a major candidate for biological warfare. As early as the French and Indian War, smallpox was used as a biological weapon whereby the British employed the virus against Native Americans they were battling. Currently, smallpox is considered one of the best candidates for biological warfare, not only because of the historical setting in which it can annihilate human populations effectively, but also because of its own characteristics as a pathogen. It infects only humans, spreads rapidly, and has a mortality rate approaching that of Ebola. It has a thirty percent mortality rate in unvaccinated individuals who are exposed. An aerosol release would be the most efficacious means of dispersal because of the stability of the orthopoxviruses and the vast expanses it could cover. Also, the necessary dose for infection is quite small and the virions can survive over 24 hours airborne as long as they are not subjected to heavy ultraviolet light exposure. (Henderson *et al.* 1999)

Since smallpox is believed to have developed from the closely related infectious virus, cowpox, it is foreseeable that smallpox could redevelop from these familial viruses, such as monkeypox. If mutated to fit human hosts as a successful infectious agent, these mutated cowpox or monkeypox strains could constitute new stocks of smallpox or a second-generation form of smallpox which can be used as a biological weapon (Bazin 2000).

In light of recent events of worldwide terrorism, biological warfare has entered the public domain and is of concern to the general population. Potential stockpiles in Russia and the United States may fall into the wrong hands because of unexpected turmoil or breaches of security.

As the world is undergoing anthrax scares and actual infections as terrorist actions, the use of smallpox as a biological warfare agent is not impossible. This has brought smallpox into the news as well as scientific forums in recent times. Most importantly, it is essential for people to understand the basic facts about the virus and the corresponding disease it causes. Although it will be difficult for smallpox to emerge since the only known samples are well isolated and secured, it would be a weapon nearly impossible to contain or combat before it infects and exterminates thousands, if not millions, of people (Gani and Leach 2001; O'Toole 1999).

There is no way to tally up exactly how many people have lost lives and suffered losses due to smallpox infections throughout history. Since then, the World Health Organization has worked to eradicate smallpox, which was a great success. It took over ten years and countless dollars to eradicate smallpox completely and now it has set the scene for a possibility of reintroduction of the virus as a competitive biological weapon. Some people argue that smallpox in secure stores must also be destroyed in order to eliminate the possibility of such biological warfare. If the smallpox escapes the secure protection and research

organizations where it is used for scientific study, it would be a disaster because of the susceptibility of the global population.

The reason that smallpox was so successful as a human pathogen is that it apparently has several proteins that interact with and disable the human immune system. Many of the new viruses such as Ebola and HIV also destroy the immune system. A critical study of the smallpox virus and how it interacts with the human body has the potential to provide valuable information that can be used in the study of other diseases. In addition to that rationale, many people are

advocates of keeping secure stocks of smallpox for research purposes. The ideal rationalization for this is to be prepared for any phenomena presented by nature. Natural selection can recreate smallpox from its ancestral relatives such as cowpox and monkey-pox. Also, as a biological weapon, we would need the vaccines and more modern methods of prevention and combating misuse of smallpox or other such viruses. Thus the overall idea in the scientific community is to keep smallpox as a research tool without destroying the entire line, but it should be kept as a secure research tool and not be given the opportunity to reemerge into society.

## Defining Disability

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When asked whether the Australian government should support embryonic stem cell research, actor and disability rights advocate Christopher Reeve responded that “the purpose of government, really, in a free society, is to do the greatest good for the greatest number of people. And that question should always be in the forefront of legislators' minds” (Reeve 2002a). A number of people who heard this statement felt that Christopher Reeve – who is confined to a wheelchair due to paralysis – was promoting a utilitarian agenda that is at odds with that of disability rights advocates.

Dr. Christopher Newell points out that by arguing for the greatest good for the greatest number of people, the disabled minority on the fringes of society are disadvantaged and are stripped of their rights by the majority (Newell 2002). While Christopher Reeve later clarified that his original intent was to “to remind legislators of their obligation to ensure that no law-abiding citizen is marginalized or left behind” (Reeve 2002b), the furor over his statement highlights the task of defining disability that disabled rights advocates such as Dr. Newell are struggling with.

The utilitarian agenda portrays disability as a deviation from the average mental and physical condition found in society that falls outside the normal range of human variation. Peter Singer, a bioethicist at Princeton, advocates abortion and infanticide on the grounds of disability. He argues that such actions are justified because they reduce the suffering that disabled children and their families would otherwise endure, and that by removing these children from society, more resources will be available to the majority (Khuse and Singer 1985).

In a utilitarian world, it is society that creates this suffering by imposing certain expectations upon its members. The suffering arises from the inability of the disabled person to conform to the social norm defined by such a utilitarian society. As an aberration they are excluded from social participation and must endure

stigmatization and marginalization because their abilities are perceived as inferior. By arguing that disability falls outside the normal range of human variation, proponents feel justified in the exclusion of disabled members from business and social activities because they do not meet the requirements for participation.

If we discard the utilitarian concept that disability falls outside the range of normal human variation, and instead consider disabled people to be representative of human biodiversity as Schriener and Scotch (2001) suggest, then the term disability no longer suggests that the individual is impaired. Those who adopt this view define disability as:

“the systematic mismatch between physical and mental attributes of individuals and the present (but not the potential) ability of social institutions to accommodate those attributes” [Schriener and Scotch 2001]

By redefining disability, the term no longer represents a permanent flaw of the individual, but rather a condition that can be corrected by society. This paradigm suggests that the social experience of disabled people is similar to that of other minority groups, such as Native Americans or African Americans, in the United States. Hahn (1985) demonstrates that disabled people experience exclusion, isolation, and stigmatization, much like members of ethnic minority groups have in the past.

This approach demonstrates that the assumptions of normalcy that are built into cultural practices become inadequate as they arbitrarily limit social participation. Institutions have made some progress in attempting to correct these social inequalities by providing interpreters for deaf people, Braille lettering for the blind, and wheelchair ramps for the handicapped. In these situations where the accommodations are adequate to allow the individual full social experience, then they no longer can be characterized as handicapped or disabled.

The beauty of using human variation as a model is that the definition of disability allows for a vast range of experiences and can be applied in some cases to those whose physical and mental abilities fall close to the average of the majority of

society. Consider the situation of a hearing person who is dining with a small group of Deaf people. Unless the hearing person is fluent in American Sign Language, he will be at a disadvantage and will not be able to fully participate and interact with the other Deaf people, all of whom are chatting silently.

A major goal of the disability rights movement is to increase the ability of disabled people to participate in society. Fuller participation in society requires the removal of barriers that limit the individual's ability to interact with others, and can be achieved through two avenues: the development of medical technology to alleviate the condition, and by reexamining standard assumptions of human variation underlying cultural practices.

One concern with the development of medical technology is that such development proceeds under the assumption that disability is a clinical disorder and falls outside the range of human variation. Such an assumption appears to be at odds with the human variation model of disability. As long as researchers and health care professionals continue to treat disabilities as an intrinsic flaw of the individual, then attempts to develop and use medical technology as treatments for these disorders will remain ineffective.

An alternate view is to treat this medical technology as a means that allows the individual to function better in mainstream society without changing their individual identity. Rather than using the technology as an attempt to correct the condition, the device or treatment should be viewed as a way of improving the person's potential for social interaction.

Cochlear implants, which provide the recipient with the partial ability to hear, are increasingly being implanted in young children. While this practice is acceptable since early implantation provides the child with the auditory stimulus required for maximum benefit from the implant, the child still retains the option to disable the implant later in life if they choose to do so. However, too many parents and health care professionals treat the cochlear implant as a device which corrects deafness, and treat the child as they would treat one that could hear (Christiansen and Leigh 2002). The child is expected to learn how to interact with a hearing society without any accommodations normally provided to deaf people. This failure to accept the child's deafness even while providing him with the benefits of an implant is an implicit refusal to acknowledge that the child's deafness falls within the normal spectrum of human variation.

Some parents perceive the cochlear implant as an aid, as one mother wanted for her child:

"If he could hear a car coming, and he's riding his bike we felt that it was just another option for him – we never planned on him becoming only oral ... We just wanted to make ... his life with other people outside of school and his immediate family a little easier." [Christiansen and Leigh 2002]

This balanced approach allows the child to retain his dignity as a deaf individual and participate in the Deaf

culture while benefiting from the auditory feedback from the cochlear implant which supplements the other accommodations he receives.

It is crucial to teach the child some form of manual communication such as Cued Speech or American Sign Language so that he will not have to rely solely on the information he receives from his implant. If this child is brought up orally, then he will be forced to rely on lipreading and the auditory cues provided by his implant. While it is difficult to distinguish between ambiguous sounds when using lipreading alone, the auditory feedback provided by the cochlear implant can provide the necessary level of information required to distinguish between these ambiguous sounds. However, many implant users report that this system fails in noisy environments or in meetings where several people are talking at once. By recognizing these limits and being able to use both an interpreter and his implant, his disadvantage in mainstream society will be minimized.

Cochlear implants illustrate the unique situation that many disabled people find themselves in. They have access to technology that mitigates their disability; but such technology is not sufficient for full interaction and they remain at a disadvantage in mainstream society. Societal assumptions must be reexamined to eliminate or minimize this disadvantage by providing appropriate accommodations.

Will there ever be a world where all buildings and locations are readily accessible to the physically handicapped, all movies and television shows captioned, and all signs Braille-lettered? There has been significant progress towards the goal of equal access for all groups. A few years ago it would have been almost impossible to obtain health insurance coverage for cochlear implant surgery, many locations were inaccessible by wheelchair, and most signs did not have Braille lettering. However, in the past few years there have been successful lawsuits against insurance providers that demonstrate that a failure to provide coverage for cochlear implants is discrimination against the disabled, and a number of places have now installed Braille signs and wheelchair access ramps.

There still remain many limitations that affect the ability of disabled people to participate in society. While the employing agency might be willing to hire the individual and provide various accommodations as required by the ADA to avoid discrimination, the employee still remains at a disadvantage in the workplace because he requires certain accommodations to be able to interact with his coworkers and accomplish his job. However, Schrinier and Scotch (2001) argue that if the ADA is remodeled after the human variation paradigm, then the workplace needs to restructure how it functions to accommodate the employee's needs.

Even if the government adopts the human variation model of disability in favor of the current civil rights model – which fails to recognize the various and unique needs of different disabled individuals – advocacy and legal and social pressure will still be needed force people to reexamine the standard assumptions underlying their cultural practices. Continued efforts are needed to encourage employers and other agencies to provide the necessary accommodations required for disabled people. As long as society continues to exclude a person from normal interaction, disability will continue to exist.

# Essays

## Neandertals' Role in Hominid Evolution

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*Key Words:* Neandertal, Cro-Magnon, human evolution, Hominids

Since the very first discovery in the Neander Valley, Germany, back in 1856, Neandertal man has occupied a special place in our taxonomic history. Among scholars, however, that special place is greatly contested. Neandertal man is the proverbial misfit when it comes to placement, not quite fitting into any model. This is the result of a lack of physical evidence, as well as conflicting research and interpretation of results. Unfortunately, as more research is completed, the picture of Neandertals' obscure placement hasn't cleared up as fast as many would like. There needs to be more information, yes, but the information available needs to be understood first. The aim of this paper, then, is to simplify, organize, and make sense of some of the data, in an attempt to understand where Neandertals really stand with respect to hominid evolution.

### *Hypotheses of Spread*

To understand where Neandertal man fits into our ancestral lineage, it will help to first understand the major hypotheses pertaining to human spread, and how exactly Neandertal man fits into both. There have recently been two emerging theories, the Out of Africa hypothesis and the multiregional or Parallel hypothesis.

The Out of Africa Hypothesis posits that all modern humans originated in Africa, and subsequently spread out over the Old World. Archaic humans living outside Africa, including Neandertal man, were all displaced by these new humans (Wong 2001). In the hypothesis, *Homo erectus*, early archaic man, leaves Africa between 1 and 2 mya and travels through the Near East. By about 0.5 mya he reaches Europe, and this is where he evolves into Neandertal man (Brown 2002). Meanwhile in Africa *Homo erectus*, in no certain terms, evolves into modern man, *Homo sapiens*, or Cro-Magnon. Sometime between 100,000 to 50,000 years ago Cro-Magnon left Africa and spreads across Europe and Asia. Somewhere along the way, or certainly upon reaching Europe, he will encounter Neandertal man. During the next few thousand years it is unclear whether Neandertal man and Cro-Magnon have any contact or interbreed, but they are both living in the same environment (Brown 2002). Those who believe the

Out of Africa Hypothesis generally also believe there was no interaction or gene exchange between populations. Within a very short time of Cro-Magnon showing up, however, Neandertal man dies out as recently as 28,000 years ago, possibly as a result of direct competition with Cro-Magnon or un-favored natural selection in the face of climactic change (Stegmann *et al.* 2002). It is now recognized that the extinction occurred gradually when compared to the arrival of modern humans. This gradual extinction supports one theory that Neandertals, because of the climate change, faced slightly negative selective pressures, while Cro-Magnons were not affected. This resulted in slightly higher reproductive success for Cro-Magnon, and is why they outlived the Neandertals (Stegmann *et al.* 2002).

The second hypothesis is known as the multiregional or Parallel hypothesis. Its main premise is that all modern humans evolved from one ancestor, *Homo erectus*, and are the result of a convergent evolution after leaving Africa (Brown 2002). It begins with the same group of *Homo erectus* humans leaving Africa between 1 and 2 mya. The individuals leaving Africa then spread, populating areas of Europe, Asia, and Indonesia, via the Near East. Initially alike, the separate populations diverge, both genetically and morphologically, as a result of different environmental conditions (Relethford 2001). However, because of similar natural forces, and groups that interacted and interbred, they do not diverge enough to become separate species. Instead, each semi-isolated group of archaic humans ends up as distinct and uniquely different populations of modern humans, *Homo sapiens* (Brown 2002).

Modern humans then, according to this hypothesis, evolved from ancestors in their own respective environment, as opposed to evolving in Africa and then spreading out. The populations evolved in multiple regions in parallel with one another.

Neandertal man's implication in the hypothesis is this: if modern Europeans evolved from their regional ancestors, they should be more closely related to Neandertals than any other ancestral *Homo* species. This is because Neandertal man occupied much of Europe, and would arguably be the one who evolved into modern day European *Homo sapiens*.

There is another hypothesis that attempts to combine logical aspects of the previous two. It is often called the Weak Out of Africa Hypothesis. Proponents of this hypothesis agree that

*Homo sapiens* left Africa, spreading across the old world, but that they did not evolve into modern humans outside Africa. Given current diversity levels, it is estimated that for most of history our population has been relatively small, about 10,000 people. With such a small population it would have been impossible for archaic man, stretched across the entire old world, to remain in contact with other groups frequently enough to facilitate gene flow and so remain as a single species. They argue instead that once modern man emerged in Africa, he then split into smaller diverse populations. These small populations independently emigrated from Africa and spread throughout Eurasia over the same time period (Brown 2002). This hypothesis takes the middle ground in this debate by trying to combine features of both hypotheses into one.

#### *Where Does Neandertal Fit?*

Given the information above, there are basically three ways Neandertal can fit into the scheme of hominid evolution. Modern Europeans can be viewed as completely independent of Neandertal. One theory that, if true, will support this claim is Cro-Magnon out-competing the Neandertals. It can be argued that physical competition was going on, or simply that Cro-Magnon had superior technology, social skills, and higher learning abilities than Neandertal. As a result of the competition between species, Cro-Magnon either killed off Neandertal, or simply outlived him because he was better adapted to the environment (Brown 2002).

The second theory suggests that modern humans have been genetically influenced by Neandertal man, but to what extent is not known. A recent area of intense debate is whether Cro-Magnon and Neandertal possibly interbred. Most scholars believe that interbreeding did not take place; however there is also evidence to the contrary. It's possible that due to positive selective pressures on Cro-Magnon genes and negative selective pressures on Neandertal genes, eventually most Neandertal genes are wiped out within the population. This leaves modern Europeans slightly related to Neandertal man, but lacking the distinct genetic variation to prove it (Brown 2002).

A third possibility is that modern Europeans are related to Neandertal man as a direct descendant, *de facto* supporting the multiregional or Parallel evolution hypothesis. Studies supporting this claim often show that certain populations, such as Asians and Australians, display very similar characteristics as their ancient ancestors. The argument is that those characteristics would not be present had the modern populations not evolved directly from those regional ancestors (Pearson 2000).

There are any number of taxonomic trees, cladograms, and other ancestral trees to help visualize where Neandertal falls in the hominid timeline. Each is arranged slightly differently, so after a short while it becomes increasingly difficult to understand why who is being placed where based on what. There are two basic kinds of evidence used in determining where a species might fit

into the scheme of evolution: physical remains and genetics. We will look at both.

Classification of organisms based on fossil evidence runs to the very core of anthropology. Fossils are pictures of the past, and can provide scientists with a great deal of information.

There are many fossil remains of Neandertal man. The difficulty in many cases is determining which apomorphic features between and among specimens distinguishes them as a species. In the case of Neandertals, however, there does seem to be enough good fossil evidence to separate them based on apomorphic characteristics. While there are still some discrepancies, many scholars have agreed upon some common features necessary when classifying Neandertal man (Pearson 2000).

Common characteristics used to distinguish Neandertals from modern humans include cranial morphological differences as well as skeletal differences. However, emphasis is usually placed on cranial differences rather than postcranial ones (Pearson 2000). The absence of a chin, large curved brow ridges, occipital bun, spoon shaped incisors, distinctive mandibular evidence, as well as a distinct low, wide shaped head all facilitate in identifying Neandertals (Rak *et al.* 2002).

While there are many fossil remains of Neandertal, there are relatively few good fossils of other archaic humans. This lack of fossil evidence has made comparisons and distinctions between the groups more difficult.

Most fossil evidence supports the claim that Neandertal is its own subspecies of *Homo*, but that it did not interbreed with modern Cro-Magnons (Balter 2001; Rak *et al.* 2002). There are recent claims of hybrid children from parents of Neandertal and Cro-Magnon, but the fossil and other evidence for this is still preliminary (Balter 2001).

Some believe that based on fossil morphology and genetic testing that Neandertal should actually be placed in its own genus (Ponce de Leon *et al.* 2001). This is not a widespread belief, however, as most scholars believe the variation between Neandertals and other species of *Homo* is well within the accepted levels. It is, for example, less than the variation that exists between chimp subspecies (Ward and Stringer 1997).

Fossil morphology does occasionally lend support to the multiregional hypothesis as well. Many point to the fact that apomorphic features of early modern Eurasians bear more similar traits to archaic humans than do modern humans (Brown 2002). This would indicate admixture between the groups.

With today's increasing knowledge of genetics and technological capabilities, DNA is transforming the way anthropology research is performed. Many anthropologists are trading in their hammers for lab coats, as DNA is the siren call they can't resist. It teases scientists with the possible genetic truth to our past. Just as DNA holds the blueprint for human life, it also holds the enigmatic timeline of our genetic past. Even if we can't extract the entire truth, yet, there is still much we can learn by studying DNA.

Common genetic tests used in doing this are mtDNA sequencing, and studying Y-chromosome variation. These results are then compared with other ancient and modern DNA samples, and conclusions are made based on relative variation between samples.

There are certain limits in using DNA analysis however, both in terms of the physical DNA samples as well as contemporary understanding of genetics. Water, oxygen, and heat all help to break down DNA, so analyses of fossils older than 50 kya are rare. The best DNA samples tend to come from cool, cave environments. Contamination of DNA is a constant problem. This usually occurs when foreign DNA on a sample is also replicated during a polymerase chain reaction (PCR) meant to amplify the sample DNA segment. It is then difficult to distinguish what is from the sample DNA and what is not (Brown 2002). It's also occasionally unclear whether DNA tests are meaningful. Although genetic analysis is an enormously powerful tool, it is still in its infancy. With the right equipment analysis can be easy; the difficulty is knowing what to analyze. If the analysis or interpretation of the results is wrong, unclear and perhaps wrong deductions can be made. Interpretation of data is based on assumptions, which can easily lead to erroneous results or conclusions. Nevertheless, these two genetic tests seem to be the best we have at the current time.

In Neandertal DNA analysis, mtDNA is primarily being used for comparison. While both DNA and mtDNA have been extracted from various archaic human fossils, researchers have only been successful in amplifying mtDNA. This is most likely because there are many copies of mtDNA in each cell instead of just one, increasing the likelihood of obtaining a sufficient quantity of good DNA that can be amplified. Amplification is crucial because it allows enough mtDNA to be copied to allow for proper analysis (Brown 2002).

There are however, also specific problems associated with mtDNA analysis. One problem is mutation. Mitochondrial DNA is very susceptible to mutations, which can impermissibly effect how data is interpreted. It's the contention of some that results coming from mtDNA studies can be attributed to mutations and population size rather than on actual population relationships (Brown 2002).

Mitochondrial DNA analysis is also affected by any inbreeding within a species. If there is any interbreeding of lineages within a species, this can lead to accelerated mutation rates which will distort results. Specifically, the mutations distort natural variation, rendering relatedness and age comparisons between samples useless (Brown 2002).

Another problem with mtDNA is its limited variability. Scientists consider mtDNA to code for only one gene, as opposed to thousands of genes with nuclear DNA. Mitochondrial DNA is also only inherited through females. Male genetic diversity is not encompassed within mtDNA. For these reasons it might not be an accurate representation of our genetic makeup (Brown 2002).

To date, there have been many studies involving modern human mtDNA analysis. These studies trace human lineages based on the number of mutations within the mtDNA.

There have been far fewer studies involving archaic

human fossils. However, recently there have been three separate mtDNA sequences completed on Neandertal fossils, as well as one of an aboriginal Australian. Much new information has come to light as a result of these tests.

The ancestral tests are performed by taking a small segment of the mtDNA called the control region which is known to have variation among modern human populations and amplifying it using PCR. The amplified mtDNA can then be compared to the same segment of mtDNA from modern humans, other archaic humans, Neandertals, chimps, or other organisms. The number of unique base pair combinations between the two samples indicates their relatedness. In this manner it was determined that the average difference between Neandertal mtDNA and modern human mtDNA is between 20 to 30 base pairs. The average difference between Neandertals is 4 base pairs, and the variability among modern humans is 6 to 8 base pairs (Kriings *et al.* 1997; Ward and Stringer 1997).

From this analysis, and the known variation between modern humans and chimps, and when our most common ancestor lived, it is estimated that Neandertals and humans diverged between 850 and 350 kya (Ovchinnikov *et al.* 2000). This would be up to six times farther back than the 150 to 120 kya emergence of modern humans. It's also concluded that modern humans share no mitochondrial DNA with Neandertal man, noting however that this does not rule out the possibility that nuclear DNA is shared (Brown 2002). The data also show that modern Europeans do not share a greater proportion of genes with Neandertal man than other populations of modern humans (Relethford 2001). This would seem to support the Out of Africa hypothesis.

Scientists have recently completed the mtDNA sequence of an archaic human from Australia (LM3), which lends support to the multiregional hypothesis. It was discovered that the fossil remains of this early human contain an ancestral root form of DNA that is different from both Neandertal and modern human DNA, which predates all modern day DNA lineages (Relethford 2001). The question then becomes how does this 66 kyo individual in Australia who contains older DNA than Cro-Magnon end up in Australia appearing to be a primitive Australian. Those who believe the multiregional hypothesis of spread point to this as proof they are correct.

There is another genetic test, which uses the Y-chromosome to test between sample variations. Where mitochondrial DNA is inherited only through females, the Y-chromosome is only inherited through males. As a result, this again may not be a completely accurate test, but it has one advantage over mtDNA analysis; it is much less susceptible to mutation. It should therefore give a more accurate picture of our genetic past. Results of Y-chromosome analysis tend to agree with results from mtDNA analysis, adding further evidence for the Out of Africa hypothesis (Brown 2002).

An apparent problem with using both mtDNA sequencing and Y chromosome analysis is natural selection. Natural selection effects mtDNA and the Y-chromosome in a very similar fashion as does population expansion and contraction. It can create the illusion of expansion or contraction, when in fact there is nothing taking place (Brown 2002). Other methods of genetic testing such as STR, STRP, STRP haploid systems, and other tests using chromosomes 1, 16, 22, and the X-

chromosome, are also affected by selective pressures (Brown 2002). Both directional and balancing selection affect results, and in different ways. Until it is determined exactly how these different selective pressures affect our genes, and consequently the data extrapolated from genetic analysis, our interpretations will continue to have limited value.

Despite the many current problems associated with DNA analysis, it does still offer a plethora of information. As analysis techniques continue to improve, and as we continue to learn how to interpret results, DNA will undoubtedly reveal many secrets of our evolution, in the process unlocking the mystery of where Neandertal fits in that evolution.

### Conclusion

Most information does point toward the Out of Africa Theory. With that in mind, there is also little evidence that Neandertal and Cro-Magnon humans actually exchanged genes. This has led many people to believe Neandertal was replaced, one way or another, by Cro-Magnons when they arrived in Europe.

However, the available information is illusive and confusing. Often, the same data is used to support different theories depending on how it is interpreted. It's easy to collect information and use statistics to create artificial relationships. Even if good data can be collected there might be uncertainty as to whether it was processed correctly and without bias. There are variables that are hard to control when dealing with genetic information. There are also, undoubtedly, variables that are not yet known that could be affecting data. This leads to data, while good, that may not be interpreted correctly. There is often a lag between learning information and understanding information. Until scientists fully understand the information they obtain, there will only be limited advances. For example, there has been a re-

analysis of data often used by supporters of the Out of Africa hypothesis to disprove the multiregional hypothesis. The new analysis shows that data used in disproving the multiregional hypothesis was interpreted wrong. While the new analysis does not show an affinity towards either hypothesis, more importantly it does not rule out the possibility of multiregional evolution, as it had been interpreted to do, based on variation analysis.

These issues surrounding Neandertal man abound. For the time being it is inconclusive where Neandertal man actually fits into our ancestral tree. Indeed, we know much more now than we ever have, but as indicated, much more is needed. We also must know how to correctly and soundly interpret that data, which is often much more difficult than obtaining it.

As more information becomes available, it will likely continue to support both the multiregional and Out of Africa hypotheses. This will lead to the two hypotheses becoming more alike, as the Weak Out of Africa hypothesis already indicates. As this happens, it statistically becomes harder to prove one theory correct over the other. This will lead to a merge of the two hypotheses, in some kind of a Parallel Out of Africa Theory.

Most evidence will continue to support the idea that Neandertal is an entirely separate species of *Homo* who didn't interbreed with Cro-Magnons; however there will also be mounting evidence (maybe not specifically with Neandertals) that supports direct, regional, ancestral evolutionary relationships. This will lead to a convergence of the two theories, where the presence of some genetic relationship is agreed upon, but to what extent is variable or unknown, especially with respect to Neandertals.

Although it may not be certain where Neandertal fits at the moment, there is certainly more information available than in the past. As the reams of information are analyzed, Neandertals' role in hominid evolution will be increasingly evident. It is only through this process that Neandertal man will be completely and correctly understood.

## The Universality of Facial Expressions across Cultures

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*Key Words:* non-verbal communication, body language, facial expression

Social animals such as human beings must communicate a wide variety of information, both emotional and factual, and to do this humans employ a wide variety of communicative signals. Facial expressions are some of the most subtle but powerful and versatile signals. Humans use these signals in a variety of social contexts including cooperative and competitive interactions, mating, and child-raising. These signals are an interesting mix of spontaneous production and intentional information transfer, meaning they reveal a relationship between human biological and cultural factors. Therefore, an analysis of facial expressions

and their usage across cultures can provide valuable insight into the extent that fundamental human behavior is culturally influenced.

The first major biologist to turn his attention to facial expressions and their significance was Charles Darwin, who wrote *Expression of the Emotions in Man and Animals* in 1872. After the writing of this book, results in this area seemed to be conflicting and serious progress in understanding facial expressions would not be achieved until the late 1970's and early 1980's, when the field was catalyzed by Dr. Paul Ekman. Ekman's pioneering work was

the discovery that facial expressions and emotions, or at least their physiological indicators, can affect each other. In addition to this, Ekman catalogued and categorized facial expressions, as well as demonstrating that facial expressions are mostly universal across cultures (Ekman 1999).

A major question for researchers of human facial expressions has been to establish their universality. This has important implications for the analysis of facial expressions because, if the expressions are found to have the same significance and structure across races and cultures, it can safely be assumed that the biological structures and mechanisms underlying these expressions are shared as well and also that they might be present in other, related species. This gives greater significance to facial expression research because the expressions cannot be considered artifacts of culture, and they can also possibly be studied in an evolutionary context.

The first researcher to attempt to establish the universality of facial expressions was Charles Darwin. Darwin reported in 1872 that he had conducted a survey of individuals of many different ethnicities distributed throughout the world and found that emotions and facial expressions corresponded in much the same way they did in his native land of England. This research, however, was not up to the standards of modern science, as he asked leading questions and often relied upon second-hand sources for his information. Nevertheless, this view was generally accepted until the 1940's.

The first of the major challenges to Darwin's view was raised by Otto Klineberg, at the time a famous social psychologist. Drawing on the research of anthropologists, Klineberg formed the view that only a few emotional expressions, such as crying, trembling and laughing, have universal meaning, many others do not. Klineberg also attacked the evolutionary implication of universality in human facial expressions, claiming that humans could not correctly understand chimpanzee facial expressions. Challenges of this sort continued to gain strength until the 1970's, when anthropologist Ray Birdwhistell noted that individuals from different cultures smiled in various positive and negative situations, seemingly dispelling any idea that such expressions had universal meaning.

The primary response to these challenges came from Ekman, who revived a claim made originally in 1862 by Duchenne de Boulogne that there were actually physically different types of smiles, with different meanings (Ekman *et al.* 1988). This distinction explained why some individuals were smiling when they were not happy. In response to the claim that human beings do not have facial expressions analogous to those of chimps, Ekman and Chevalier-Skolnikoff, a primatologist, found that information to be misguided. The research that Klineberg used to establish a perception gap between chimpanzees and humans was conducted by comparing the emotional analysis of the photographer who took the primate pictures with students' attributions of emotion to the various expressions. When the students' results were compared with Skolnikoff's expert analysis of the expressions, rather than the photographers, they were found to be correct (Ekman 1999).

The most fundamental and versatile of human facial expressions is the smile. Ekman classifies smiles into several different categories, but the most important distinction he makes is the difference between a true smile, also called a "felt happy" or "enjoyment" smile, and the false smiles. The enjoyment smile is produced spontaneously when an individual feels happiness and is differentiated from all other smiles by the contraction of the outer part of the orbicularis oculi, a muscle around the eye that Ekman found is rarely able to be moved intentionally. False smiles are made to convince others that a positive emotion is being felt, when in fact it is not. There are two types of false smiles: masking smiles and phony smiles. Masking smiles are produced to cover up a negative emotion with a positive one, while phony smiles are produced when no particular emotion is felt.

Using this set of definitions, Ekman was able to make predictions about which smiles would be produced in which contexts, providing a theoretical basis for testing. The most obvious of these predictions is that subjects would produce enjoyment smiles more often than false smiles when describing events from the past which they enjoyed, and that the opposite effect would be found when subjects were describing events about which they had negative feelings. Phony smiles should look the same as enjoyment smiles, except for the muscle around the eye that will not be contracted. Masking smiles will appear as some combination of the facial expression associated with the emotion the subject is trying to mask and a phony smile (Ekman 1992; Ekman 1993; Ekman 1999).

Ekman verified these predictions in a 1988 study in which volunteers were told to describe an event they found pleasant, and also to describe watching a disturbing video of a gruesome surgical process as if they very much enjoyed it. Ekman found that those describing pleasant events produced enjoyment smiles often, while those lying about their experience of watching the video produced almost entirely masking smiles. Independent verification of the distinction between enjoyment smiles and false smiles comes from studies such as Schneider's 1987 study showing that five to seven-year-old children produced more enjoyment smiles after winning a game than losing it.

This discovery has important implications for the universality of facial expressions, as it shows previous findings that smiling rate was not related to emotion to be misguided. Ekman found that, when he considered all types of smiling without making this distinction, he also found no correlation between the amount of smiling and the honesty or emotional content of the subject. Thus, using smiling alone as an indicator of the subject's emotional state yields confusing results, but using only enjoyment smiles as a standard for comparison yields clearer results.

Ekman and his colleagues also emphasize the connection between the facial expression associated with an emotion and its physiological effects. In addition to the common understanding that emotions create facial expressions, they have performed studies which indicate that the production of a facial expression produces the physiology characteristic of the emotion associated with the expression. In one such

study, Ekman *et al.* (1988) monitored many physiological parameters such as heart rate, finger temperature, skin conductance, and autonomic nervous system (ANS) activity of subjects asked to perform certain facial expressions. These subjects were told not to make the expression or to feel the emotion, but rather were read a detailed, muscle-by-muscle description of the expression and told to perform it, both with feedback and without it, to control for reaction to seeing their own expressions.

The researchers found increased heart rate and increased skin conductance associated with the production of anger, disgust, and fear expressions than for happiness expressions. Finger temperature was greater for anger than for fear. With regard to ANS activity, the same patterns when producing a certain expression were found not only among members of the original test groups, but also among older individuals, and people from a culture differing from western society in almost all defining aspects, including religion, language, and social organization, the Minangkabau (Ekman 1992; Ekman 1993; Hauser 1996). Combined with Ekman and Levenson's other results, these further rule out the possibility that facial expressions are an artifact of a particular culture, showing that not only do members of other cultures produce the same spontaneous expressions, when properly defined, but also that they experience the same physiological effects during their production.

One of the major challengers of Ekman's work has been Fridlund, who examined facial expressions with more emphasis on social interactions, as opposed to their internal effects and mechanical production. Fridlund poses two claims as challenges to the view that facial expressions are caused by certain emotions: that all facial expressions are made to be viewed by some group of people, real or imagined, and that facial expressions have evolved to manipulate others, rather than merely conveying information about emotional state. Thus, facial expressions would almost always contain some deceptive aspects. While this is not in direct contradiction to the traditional relationship between facial expressions and emotions, it is more complex (Hauser 1996).

To test his view, Fridlund used mental imaging techniques to study subjects watching a comical videotape under one of several different conditions. First, the subject comes to the test alone and views the tape alone. Second, the subject comes to the test with a friend and watches the tape alone, but is told that the friend is being tested in some other way. Third, the subject arrives with a friend and views the tape alone, but is told that their friend is watching the video as well. Fourth, the subject and friend watch the tape together in the same room. Fridlund found a significant increase in smiling between the first and fourth scenarios, but no difference between the third and fourth tests (Hauser 1996).

From these results, Fridlund concluded that the presence of other people, whether real or imagined, can have a significant impact on the production of facial expressions. Fridlund then argues that the established correlations between ANS change and the production of facial expressions is not enough to tell whether this correlation is

related to a change in emotion, rather than simply display function. Fridlund, therefore, claims that facial expressions can be understood without reference to emotion, which seems to conflict with Ekman's view. However, it is important to note that Ekman never claims that facial expressions cannot be used intentionally or deceptively, in fact the performance of such expressions is at the core of many of his tests. The conflict between Fridlund and Ekman seems puzzling because they are both essentially talking past each other, with Fridlund attempting to describe the function of facial expressions in a social context, and Ekman attempting to address the mechanisms of their production. These two understandings are not mutually exclusive, and will both be important in a final understanding of facial expressions (Hauser 1996; Keltner and Ekman 1996).

With facial expressions providing such a potentially clear indicator of emotional state, it would be expected that human beings, engaging in social activity often, would have a good deal of success at assessing these states, especially when this facial information might be combined with other cues. In any communication, one of the most important factors to determine is whether the data gathered is reliable, in this case, whether the other person is lying. Given this and the success of Ekman's studies, it seems to follow that humans would be excellent lie detectors. However, this does not appear to be the case. In a series of studies of studies by Ekman *et al.* (1988), subjects under a variety of circumstances failed to identify liars at a better than chance rate. These studies ranged from low to high-stakes lies, reward and punishment scenarios, situations where the subjects were free to choose whether to lie or not, and experiments testing different groups of people for their lie detection ability. Of these, the only group found to detect lies at a higher than chance rate was secret service agents, which were found to detect liars at a 73% rate (Ekman 1996; Ekman and O'Sullivan 1991). This anomalous result is ascribed to vast experience with lie detection in high-stakes situations, not a natural talent or ability.

These results force a consideration of deceptive facial expressions in an evolutionary context. First, it would seemingly not benefit an organism to evolve the capability to deceive other members of its species, and then also to produce reliable cues whenever it is doing this. Further, if such clues to the truthfulness of another's expressions exist and are detectable, it would follow that an individual born with a greater ability to detect these clues, or at least a natural disposition to disbelieve those giving off such indicators of deceit. There are two parties in this situation with conflicting interests: deceivers, who want their false signals to go unnoticed by others, so they can reap the benefits of deception, and perceivers, who want to determine whether others are presenting information to them truthfully. This scenario resembles the classical setting for an evolutionary arms race.

However, Ekman doubted this scenario, and so ventured to Papua New Guinea to study what was then a pre-literate culture with technological levels similar to those found in the Stone Age. This culture was considered by anthropologists to be one of the best living models of the

social environment in which the human mind and its behaviors might have been shaped. Ekman made several observations about the way lying functions in their society (Ekman 1992; Ekman 1997).

The most striking contrast between this culture and modern ones, as reported by Ekman, is that they have very little privacy. They live in closely-knit, small communities with much social interaction. In this climate, members of this society did not attempt to detect lying by any analysis of expressions or other clues, but rather by finding factual evidence that the lies were false. For instance, adulterers were most often caught by being found hiding together somewhere on the outskirts of the village. Also, lying often had great consequences for the liar if he was found out. Since much of the hunting and gathering of food was conducted cooperatively, losing the trust of other group

members would have disastrous consequences. Also, the lack of social mobility, because various tribes were isolated and the variety of jobs was not great, meant that a liar, once branded, had little or no ability to escape the consequences. Further, such large social pressure against lying would have been enough to keep its incidence low, and thus lie detection never became a skill essential for survival, which would have motivated its development by natural selection.

The evidence presented by Ekman and others indicates strongly that human facial expressions are produced under similar circumstances by peoples of all cultures. Even in something as complex, personal, and socially driven as facial expressions, the underlying biological unity shared by all human beings can be seen. Results such as this one show that science can confirm and strengthen man's fundamental convictions about equality, and even do it with a smile.

## Ellis-van Creveld's Syndrome in the Amish

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*Key Words:* Lancaster County Pennsylvania, Chondroectodermal dysplasia

Living in Lancaster, PA for eleven years, I have grown up being accustomed to seeing the Amish around. Usually they are in the slow buggies that I am stuck behind when traffic is coming in the opposite direction and I need to be somewhere. For me, the tourist hype over the Amish seems odd. The Amish come into the places I work and they seem like any other person to me. They are customers like anyone else. It seems to me in this case that a lot of people look on with caution because they're beliefs and practices are so different. Even though I have grown up around them, and am so familiar with them, I realize that some of my perceptions of the Amish are misled. I always thought that Ellis-van Creveld's Syndrome, which has a high prevalence among the Amish, was a result of inbreeding, rather than being caused by an evolutionary bottleneck. Before I discuss Ellis-van Creveld's Syndrome I will give some background information on the Amish.

The Lancaster County Amish are descendents of about 200 Swiss people who came to America in the mid 1700's. The Amish are a very conservative Christian group. Their beliefs are similar to those of the Fundamentalists. They believe in a literal translation of the Bible. They live in a community by themselves, separate from the rest of the world. When I say separate I mean they mostly keep to themselves. They have their own communities but that's not to say I don't see them at WaWa's [ed. Note – WaWa's is a convenience store chain that is popular among University of Maryland students] just like other people. They shop at Walmart, Rite Aid and other stores. The Amish are opposed to military involvement and warfare.

Each Amish community is autonomous. There is no

centralized head of the Amish organization. The Amish do not seek converts. In that respect they are not like other Fundamentalist churches. Members of the Amish community do not own or operate automobiles. Yet, there is an alternative to going somewhere in a buggy. The Amish cannot drive automobiles but they can pay someone to drive them around. The Amish also do not use electricity, so they do not have televisions, radios or computers in their homes.

Members of the Amish community are not allowed to marry anyone outside the community. Amish marriage ceremonies are usually held in December after the harvest season. Amish dress is very plain. Both men and women wear plain suits and dresses in solid colors. Amish religious services are held twice a week. The services are held in a different family's home each week. Amish funerals are very simple. There are no flowers or decorations. The casket is plain and there is no eulogy said.

The Amish collect money for mutual aid funds for the whole community. The money can go toward medical cost, dental bills or other similar things. Typically, Amish children do not attend formal schooling after the eighth grade. The Amish are a bit of a tourist attraction in Lancaster County. There are many cheap touristy places and buggy ride tours people can visit. The Amish are sometimes extorted. The Amish community does not take photographs. Funny how you can find postcards with the "Amish" pictured on it (Robinson 2002).

Ellis-van Creveld's syndrome, also called Chondroectodermal dysplasia, "is an inherited disorder with multiple abnormalities and dwarfism" (Brooks 2001). "The name chondroectodermal is used because the condition affects the

skeleton (chondro-) and the skin (ectoderm)” (Greenberg 2002). It was originally described by Richard Ellis and Simon Van Creveld in 1940. Ellis-van Creveld’s syndrome is a recessive autosomal trait. Degrees of severity differ from person to person. Although Ellis-van Creveld’s syndrome is not limited only to the Lancaster County Amish, it is most prevalent in this community. Ellis-van Creveld’s syndrome is characterized by short limbs and postaxial polydactyly which is when a person has a sixth finger on each hand next to their pinkie finger. I knew dwarfism was a problem among the Amish but I did not realize that these other traits were part of the same disease. This occurs in almost 100% of all people that suffer from Ellis-van Creveld’s syndrome. “In addition, 10-25% of people with [Ellis-van Creveld’s syndrome] have an additional digit on the feet” (Greenberg 2002). In most cases the extra digits are removed surgically. Congenital heart defects are a problem in about fifty percent of all Ellis-van Creveld’s syndrome patients. The most common problem being atrial septal defects, “which means there is a hole in the heart between the upper two heart chambers” (Greenberg 2002). This heart problem can be corrected in most instances surgically.

Ellis-van Creveld’s syndrome has many symptoms. Still birth and death in early infancy are common symptoms. Dwarfism is a symptom on Ellis-van Creveld’s syndrome. This includes short extremities, especially the forearms and lower legs. Along with shorter limbs the range of motion is often limited at the joints. As mentioned before extra fingers are also a symptom of Ellis-van Creveld’s syndrome. Hair abnormalities are another common symptom of this disease. It is often sparse, absent or fine textured. Under microscopes, it was seen that hair had and irregularly grooved surface lacking in scales. The sulfur level of the hair was about half of the normal content. I was really surprised when reading that their hair was not as strong. I’d always heard that the reason Amish went bald was because they pulled their hair too tight back into under their caps. This could be part of the reason but a better hypothesis may be because of Ellis-van Creveld’s syndrome. Nails are often abnormal as well with people suffering from Ellis-van Creveld’s syndrome. The nails are often deformed or in some cases missing completely. Nails were shown to break easily and do not grow as long as they should. Some oral problems also occur with Ellis-van Creveld’s syndrome. “Oral problems include frenulae or areas of fusion between the inner upper lip and gum and partial or pseudoclefts of the upper lip” (Greenberg 2002). Another symptom is teeth abnormalities. Peg teeth, widely spaced teeth, or teeth delayed or absent in forming sometimes occur.

Victor A. McKusick says in his article, *Ellis-van Creveld syndrome and the Amish*, that there are four reasons that the Amish are an interesting group to look at when it comes to medical genetics. The first being how the founder effect influences a community. As previously stated, the Amish people are all descendants of a group of 200 Swiss people that came to America. The genes and traits are limited

because the population is limited. “Second, the Amish observe strict endogamy (they marry only within the community), with the gene flow being exclusively centrifugal (that is, members may leave the community but ‘outsiders’ do not join it and thereby introduce exogenous genes)” (McKusick 2000: 203). The third reason is that they keep excellent genealogical records and tend to have a restricted geography in which they reside. The fourth and final reason they are an interesting group to study, is that they tend to have large families with many children (McKusick 2000: 203). Because the Amish keep such excellent genealogical records it is possible to trace both parents of all the Amish people that have Ellis-van Creveld’s syndrome back to one couple. The couple they can trace back to is Samuel King and his wife, who came to America in 1744. This is more proof of the founder effect working with recessive traits to make homozygous recessive traits more prevalent (McKusick 2000: 203).

Victor A. McKusick goes on to say that the medical community and genetic research owe a lot to Amish people who participated in studies. They have led to a better understanding of some genetic disorders. He says the information they know now can help when or if a couple goes to premarital and prenatal counseling. However, it is unknown if Amish would go through premarital testing. “It is unlikely that they would accept prenatal testing because of the implication of abortion” (McKusick 2000: 204). I was surprised when reading this article. I had always heard that the Amish did not like to see doctors if they did not absolutely have to. I thought genetic testing would be out of the question. McKusick says, “The Amish acceptance of the geneticists was achieved by their being introduced by local physicians and by sociologists who they trusted” (2000: 204).

After reading up on Ellis-van Creveld’s syndrome I realized that I did not know as much as I thought I did about the Amish. I heard that the Amish had genetic problems because of too much “interbreeding” like cousins marrying cousins. That idea is not quite right. Yes, the problems stem from being closely related but not necessarily as in a family relation. It is just a problem that stems from the founder effect. Because the original community started with only 200 people eventually all the genes were passed around, genetic possibilities almost exhausted. I major surprise I came across while reading this was the discovery that genetic testing had been done within the Amish community. I guess it makes sense. They had to get the information from somewhere, but I had always heard that the Amish did not usually seek medical attention or consult a doctor. I had heard stories where children had not had tetanus shots and had serious problems because they cut themselves on farm equipment. I assumed that this meant they would be against genetic testing. I realize that this information could be of some help to the Amish community though I do not see how it could possibly get better unless they start marrying people outside their communities.

# Race and Hate

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*Key Words:* Nazi Germany, social Darwinism, Holocaust

Anthropologists study issues involving humankind that should be applied for the betterment of it, but sometimes their conclusions can have terrible consequences. One of these issues that is still pervasive in the year 2002 is racism. While it is true that things are better than they were 100, or even 50, years ago, racism is still endemic. There have been many atrocious and violent acts of racism across the world and throughout history that include, but are not limited to, the colonial annihilation of Native Americans, trans-Atlantic slavery, apartheid, and genocide in countries like Rwanda, Iraq, and Serbia. One of the most egregious and insidious genocidal acts of racism, and definitely the best known, was the Holocaust. While anthropology was not solely to blame for the twisted mind of Adolph Hitler, the opinions of some anthropologists, sociologists, psychologists, and philosophers helped give him the fuel he needed to ignite a country in hate. Race is a concept that is not accepted as a biological truth by most anthropologists today, but it still holds in some sectors.

There were compounding events that led to the outbreak of World War II. The harsh treatment of Germany after World War I, its deplorable economic situation, and the egotistic religious convictions held by much of the country were a few of these overwhelming conditions, but the main reason for the Nazi movement in Germany, stated by Hitler himself, was based on race (Lightfoot 1972). Adolph Hitler's ideologies were rooted, in part, on the writings of the German anthropologists D.H. Kerler and Nicolai Hartman (Krzesinski 1945: 36). They espoused the idea of life pan-romanticism, which translates into the right to live however one wants (Krzesinski 1945: 37). Other German scholars such as Nietzsche and Kant expressed the same attitude of self-preservation and ego-power. The psychologist Leopold Ziegler said that man should dispense with God and "take His place and endow himself with all God's attributes" (Krzesinski 1945: 39). These ideologies were manifested in the countless experiments conducted on innocent humans under the auspices of the Nazi regime as well as the mass execution of non-Germans.

Hitler distorted Darwin's theory of evolution through natural selection by applying it to a social context. Darwin's theory states, in short, that those individuals who are most suited to their environment will survive, while those who are less fit will become extinct. Social Darwinism is the same basic concept, except it considers a person's social status rather than biological characteristics. Hitler went a step further to assert that social status is a biological characteristic. He defined race based on the hereditary biological and sociological characteristics (Krzesinski 1945: 39). In other words, what would be considered the German Nordic race was, in Hitler's mind, equal to the German

nation. Anything and anyone that did not originate in the German nation was inferior. According to Andrew Krzesinski, based on Hitler's infamous publication *Mein Kampf*, the underlying principle of the Nazi movement was that:

"The law of natural evolution, which governs the whole world, demands that a superior race be the master of inferior races: therefore, the German race should govern the rest...Every German is obligated devotedly to guard this racial purity...Whatever he may do to accomplish this end, or to promote German power over all the other races is good, licit, and desirable." [Krzesinski 1945: 41]

This principle is in keeping with the doctrine of German academia of that time.

One major flaw with Hitler's argument, even if it was effective, was his notion of race. Today's definition of race is "each of the major divisions of humankind, having distinct physical characteristics," or "a tribe, nation, etc., regarded as of a distinct ethnic stock" (Brace 1999; "Race" 1999). Most modern anthropologists are now arguing against race as a useful classification. The "major divisions of humankind" are historically accepted as Negroid, Caucasoid and Mongoloid (Brace 1996). These are based on skin color and physiological features and, as such, are completely erroneous. Mongoloid is used to describe people of Asian descent and assumes that all Asians generally share the same features as the Mongols. In truth the Mongol peoples are the most morphologically distinct of all the Asian groups (Brace 1996: 111). It is, therefore, completely incorrect to apply their characteristics to all people of Asian descent.

The categorization of Negroid is based solely on skin color, with negro meaning black in Spanish (Brace 1996), but the morphological differences between people of color are remarkable and numerous. From one area to another, so-called Negroids differ in nose size and shape, amount of pigmentation, stature, hair texture, and mouth size, to name but a few characteristics. The term Negroid was applied mainly to Sub-Saharan Africans, but dark-skinned peoples are found in all equatorial areas from New Guinea to South America. This supports the hypothesis of skin color by natural selection. Research has shown that higher levels of melanin block the penetration of UV-B rays, the leading cause of skin cancer, by up to 95% (Brace 1996: 115). Therefore, the dark skin shared by many people of the world is likely an analogous trait selected for survival and not a shared hereditary characteristic.

Caucasoid is a category born of Christianity, which suggests that "whites" are the least modified of God's people and descended from Noah who landed at Mt. Ararat in the Caucasus Mountains (Brace 1996: 115). There are glaring

problems with this, starting with its politico-religious aspects. Anyone that is not Christian has good reason to immediately disregard Caucasoid as a race. Other drawbacks include arguments that have already been stated, especially the morphological diversity among “Caucasians,” to include levels of pigmentation (compare Arabs and Norwegians), stature and facial features (contrast between Europeans and Indo-Europeans).

The second part of the textbook definition of race, and probably the more commonly used, has to do with a “distinct ethnic stock.” This is more along the lines of Hitler’s racial classification since Germans and Jews would both be considered Caucasoid according to the first half of the definition. Ethnicity and race have become combined concepts, evidenced by the American categorization of African-Americans as those Americans with African pigmentation, which disregards the possibility that these same people could have originated elsewhere. African is an ethnicity. Jewish, on the other hand, is a religion that is also recognized as an ethnicity. Hitler blamed Germany’s problems on Jews and vowed to eradicate them. He believed that the Jews were trying to become “despots of the world,” starting with the Bolsheviks in Russia, and that this attitude was biologically programmed in them (Krzyszewski 1945: 85).

It is preposterous to think that actions obviously steeped in political, religious, and cultural reasoning, would be biologically programmed. Sociological characteristics, such as ethnicity, are cultural differences and have no place in biology. C. Loring Brace, an expert on race in anthropology, suggests that the most important similarity among all humans is intelligence, and that it does not differ from

region to region like “non-adaptive regional configurations of craniofacial shape and other aspects of soft-part variation” (Brace 1999). If human intelligence is the same worldwide, then there is no such thing as an inferior or superior human race. Morphological diversity is a product of environmental selection, but that same selective force required equal intelligence from culture to culture. Evidence of this is found in the rise of language 200,000 years before present (Brace 1999). Despite the different morphological features necessary for survival in different areas, selective pressures everywhere promoted the use of language. And it is language that allowed the birth of cultural variation (Brace 1999).

It is a belief in the concept of inequality that perpetuates the idea. When a group is not given the same opportunities as others that group loses out on certain benefits like education and employment. After time it begins to seem like that group truly is inferior because they do not score well on tests, have higher unemployment rates, higher crime rates, etc., but the potential of the group is the same as any other. It is the social inequality that leads to intellectual inequality (Brace 1999). German Nazis destroyed Poland and decimated the European Jewish populations. All of the hate and frustration that Germans felt about their situation was directed at innocent people who did not hold the same values and beliefs as they did. It is apparent that race does not have a place as a biological classification. Science has shown us that the human genome is 99% the same, which means that all humans, cross-culturally, are the same inside, and race is useless and incorrect. History has shown us that race is useful and correct in terms of one thing: Hatred.

## Bio-Cultural Approach to Menopause

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*Key Words:* menopause, menstruation, female, social stigma

In our heavily westernized society, the middle-aged women has long been characterized as a melodramatic, overripe, unsupportable woman irritated with, and yet in denial of, her slow downfall into spider veins, stocks of anti-wrinkle creams, and memorized excerpts from women’s self help books. A key term that is often linked to this stereotype is menopause, a word that women’s ears dread to hear and one that husbands only seem bring up with friends in conversation as water cooler jokes. At the same time, quite ironically, our society as a whole seems to be quite deficient of a true, informed understanding of this stage in human females and of the associated physical and psychological changes. Often, women that are pre-menopausal tend to view menopause as irrelevant to their lives, an issue that does not concern them, and it remains a vague concept. This, unfortunately, proves to be detrimental for when first signs of menopausal symptoms

actually do begin to manifest themselves, many do not have the sufficient background knowledge to self diagnose themselves and attribute the changes to menopause. They become uncertain and confused about what is happening to their bodies, not knowing the proper steps to take in dealing with them. Menopause thus becomes more of a shock, for “fear of the unknown.”

But why is such a natural and inevitable transition so stigmatized? Is this antagonization universal to all cultures, and does this negative attitude, in turn, affect the physiological symptoms and methods of treatment? What evolutionary purposes does menopause have to development of humankind as a whole? This essay attempts to answer the latter questions, integrating biomedical and socio-cultural approaches, while showing the symbiotic relationship between these views.

Medically, menopause is actually more difficult to measure or mark than commonly assumed. For purposes of medical

standardization, menopause is defined as the cessation of menses for 12 months. This specific labeling is already problematic, for determining the last menstruation is difficult. Frequently, during the final years nearing menopause, there is an irregularity of cycles. In addition, there may be shedding of blood not necessarily be in relation to monthly menstruation (Huffman and Myers 1999). Another, more fully encompassing way of looking at menopause is to see the stage in life as a transitional process – one not marked by a single, determining event, but by evolving change in many steps. One must also keep in mind that though menopause is a transformation that all women who live to their appropriate years must go through, it is also unique to each individual, with every woman varying in their symptoms, ways of coping, and proper methods of adjustment. “The physical event is the same everywhere, but the experience of menopause is as varied as humanity itself” (Shute 2002). The average age in the United States at which menopause occurs is 51.4 years, with 38% of women in the country being at or beyond their menopausal years, and this percent is ever growing (Gold *et al.* 2001). There is also a genetic factor that influences the age at which menopause occurs. Studies have been done showing that the age difference at which onset of menopause occurred between monozygotic twins was less than for dizygotic twins (Berger and Wenzel 2001).

Many see menopause, rather, as occurring in three biological stages. The perimenopausal stage is the period leading up to menopause. Its duration can be as long as a decade. Changes that occur in the body in this time are erratic ovulation and menstruation, hot flashes (a thermoregulatory response of the automatic nervous system known as the vasomotor aspect of menopause) head, joint, and back aches, difficulty in sleeping leading to tiredness (Brown *et al.* 2002), dizzy spells, lack of concentration, fatigue (Huffman and Myers 1999), deterioration of bone density, urinary tract infections, urinary incontinence, vaginal dryness, uterine prolapse, changes in skin texture, weight gain and dyspareunia (Al Qutob 2001). The next stage is menopause itself, followed by the third and final stage, postmenopause, where a woman stays for the rest of her life. Postmenopause sees the cessation of many of the symptoms that occur in premenopause, though some are retained and must continue to be carefully monitored.

There is much psychological change that occurs during menopause. Many attribute this to lower estrogen levels during menopause, however most psychological distress occurs during the perimenopausal stage. If the former were true, postmenopause, when there is the least estrogen production, would witness the greatest psychological worry (Bromberger *et al.* 2001). Much of the psychological component is in part due to physiological factors:

“An important concomitant of vasomotor symptoms is disrupted sleep which has also been shown to be more prevalent during perimenopause... correlat[ing] with tension, anxiety, and depressive symptoms.” [Bromberger 2001].

Another theory to explain the prevalence of psychological distress deals with the experience of life losses – such as the death of parents, health complications, children leaving the house, and escaping youth – during middle aged years and stressful situations – such as a busy workplace, taking care of elderly parents and adolescent sons or daughters – unique, but not exclusive, to this fragile time period (Huffman and Myers 1999). In most westernized societies menopause has been turned into a symbol of aging and decay. This viewpoint adds to the accumulated stress that many women already feel around their middle aged years.

An interesting phenomenon, is the impactful role that self and culture serve in influencing perception of physical menopausal symptoms. Women who anticipate a difficult transition in menopause before it occurs most often will end up having the level of pain and difficulty they predicted; this is the idea of a self-fulfilling prophecy (Elias 1993). Life style factors also contribute significantly to menopausal complications, and include factors such as educational level, ability to meet basic needs, smoking, exercise level, and body mass index (Women’s Health Weekly 2000).

Variances between global populations can therefore likely be contributed not only by genetics, but also by environmental factors such as age or ell. Nutritional value and manner of alimentation highly makes an impression on the degree of overall health of an individual. This relationship is evidenced in many Japanese women, who report having very little, if any, hot flashes, and low irritability and depression, perhaps as a consequence of the former. The exemplary diet and health behaviors of the Japanese might help explain the greater ease at which menopause is experienced, in comparison to westernized society-where hot flashes affect between 70-80% of menopausal women (Bromberger *et al.* 2001). Japanese women:

“have one of the world's longest life expectancies at 84 years [and] are more likely to eat a diet low in fat and rich in vegetables and soy, which provides phytoestrogens that may blunt menopausal symptoms, [and] get considerable exercise.” [Shute 2002]

Nevertheless, are only environmental factors and life style patterns to be credited with the people’s average low levels of menopausal discomfort? Might the fact that Japanese society places more positive, associations on aging and women’s roles, that result in greater optimism toward facing menopause, have an influence on the pain and discomfort level felt? In many non-westernized societies, respect and title accrue with age, making the elderly more esteemed individuals of their social group rather than depicting them as decrepit, sexless, un-usefuls. The Mayan women of the Yucatan achieve higher social status with age and are often alleviated of many tedious household burdens, which are passed to the daughters in law. Women, as a result, look forward to this time period where they are able to reap such benefits, and perhaps as a consequence, reports of aches and vasomotor problems are virtually unheard of (Shute 2002). This is true of many populations, including women in some African tribes, Indian women, Filipinas, and Chinese to name a few, though it must be noted that, in each, there are many individuals who do indeed report troubling afflictions that are

menopausal related. This is because, even within populations, there is still much variation from person to person.

Other socio-cultural demographic factors that are specific to developing nations have an effect on the manner in which menopause is dealt with among their people. The mortality rate is much higher in these countries, leaving less of a proportion of the population reaching or living past their menopausal years. The medicization of menopause is not nearly as prevalent as in the United States and its post-industrialized counterparts, due to scarce resources, which are mostly aimed at helping those in dire need. These situations could factor into why menopausal physical symptoms are not placed with much emphasis in these contexts (Al-Qutob 2001).

The opposite is true for most developed nations, where outlooks tend to be excessively youth oriented and pronatalist (Singer and Hunter 1999), causing women experiencing menopause to feel they are losing their beauty, admiration, competence, and “femininity” in accompaniment with their already stressing physical changes. One gynecology book from the 1920s states that menopausal women were “peevisish, irritable, morose, and depressed .... Many have full-blown insanity with melancholia, paranoia, and maniacal conditions” (Mansfield *et al.* 2000). Another quote from a 1940’s medical journal writes “The climacteric is an indication for woman that the period of her vigor is beginning to disappear forever. With more or less rapid steps but steps which admit of no return, woman now proceeds toward old age” and a later medical publication read, “After the last planned pregnancy, the uterus becomes a useless, bleeding, symptom-producing, potentially cancer-bearing organ and therefore should be removed” (Huffman and Myers 1999). Women easily acquire such pessimistic attitudes when the medical community holds such patriarchal views. Though there is more breadth to the understanding of women nearing menopause in our present day, this desensitized biomedical approach, viewing menopause as a pathological “hormonal deficiency disease” has lingered in many doctors’ views on treatment and their subsequent recommendations and prescriptions. Women find that the decision to take hormone supplements is very much promoted, almost becoming an obligatory action (Singer and Hunter 1999).

HRT stands for “hormone replacement therapy”. For women who have had their uteruses removed, taking estrogen alone is sufficient. However, with those women who still have theirs, taking this hormone alone increases the risk for endometrial cancer, thus Progestin is taken in accompaniment to protect the lining of the uterus from excessive growth and helps in combating osteoporosis, which is the brittling of the bones. Additional HRT benefits are ease of vasomotor discomforts, heightened protection against cardiovascular disease, and decline in incidence of endometrial hyperplasia and carcinoma. However, there is a trade off in choosing to take HRT, rather than letting nature take its course. One adverse effect is a possible increase in breast cancer risk, leaving

women in a catch-22 on whether they should commence HRT or let nature take its course (Elias 1993).

Indeed, the actual improvements HRT offers should be more carefully scrutinized, as the research tends to use biased sampling, with healthier than average individuals partaking in the study, and the reports of “improvements of life” having often not been sufficiently tested for true verification. Many are skeptical of HRT, stating that menopausal women are manipulated by the pharmaceutical industries who only look for economic gain, and who play on emotions and fears of women towards of aging, in making the treatment seem like an “anti-aging” solution in an effort to market their product (Gold *et al.* 2001).

Western medicinal thought tends to focus on quick solutions-targeting one specific ailment with chemicals when it is manifested. Preventive or alternative medicine is not advocated. Women need to be educated on life long living patterns, that will lead to overall quality of later years, with an infusion of mental and emotional awareness, natural herbs and remedies, and proper health and exercise measures, rather than taking five pills a day, or shots once a week.

The human species is unique in its menopausal experience. Many women live as much as a third or a half of their years in the postmenopausal stage. What does this translate to in evolutionary terms? Why is it that males are able to reproduce until death, while women are placed under the timed limits of the “biological clock”?

One trend that has been gradually occurring over the course of modern human history is that of an average increase in the duration of the human life span. This offers hints into why women live so well beyond their reproductive years. In the past, women might not have even lived long enough to witness their own menopause. Thus menopause could be viewed as a “reflection of life-history patterns adapted to a pre-technological era” (Alvarez 2000). There are many hypotheses about the nature of menopause and why humans have evolved displaying it, however, for brevity, they will only be introduced. The Risky Mother hypothesis states that due to decreased health in a woman’s later years and risk of death in child labor, it was more advantageous for women to concentrate their energies in caring for their pre-existing offspring to “enhance their genetic legacy” (Alvarez 2000). The Grandmother Hypothesis, an alternate but similar view, states that menopausal women, unable to reproduce, would devote their energies towards helping the daughter in the caring for her children. In this way, the grandmother can look after the older children so that the mother can devote her energies towards further reproduction. The Hadza people of Tanzania, a hunter-gatherer society, demonstrate this hypothesis. Studies have shown that older child, through the grandmother’s care, maintained body weight while the mother was able to breastfeed her younger offspring (Women’s Health Weekly 1997).

Adult mortality, life spans increase, and natural selection will begin to bring about characteristics that delay maturity, meaning that children will need more time and energy devoted to their upbringing. There is an inverse relationship between amount of parental care and number of offspring produced. The maturation period for humans entails a hefty devotion of

time and resources.

Knowing the evolutionary historical context of menopause in reference to human adaptation makes it evident that menopause is not an abnormality or a disease, as it has been typecast in many Westernized societies. Though many of the reasons for which menopause evolved in humans, according to the hypotheses, are not applicable today, it is imperative that women do not feel this time period to be “aberration.” Understanding the biological

processes of Menopause contextually is essential for helping women understand what is occurring in their bodies, however, more awareness of the subjectiveness and variance in symptoms cross culturally should be instilled in the greater medical community. Perhaps one culture can learn from the other in how to deal with this life transition, creating a more united and varied approach, for menopause occurs at several different levels and across several phases, none of which should be compromised.

## Gene Therapy: The Ultimate Cure?

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*Key Words:* gene therapy, Jesse Gelsinger, Ashanti DeSilva

Gene therapy is a procedure in which a patient’s genes are modified as a way to treat a genetic disease. Every feature of every organism on the planet is controlled through their genetic code and the expression of the genes themselves. When mutations occur many factors can be influenced leading to severely debilitating diseases. Gene therapy would correct the disease at the genomic level, making all other forms of treatment obsolete. Many opponents claim that gene therapy will never draw the line and soon parents will be choosing the gender as well as physical features of their children. Proponent, however, argue that gene therapy will be the end all, cure all for all the genetic diseases of the world. The patient’s body will be equipped to fight off the disease or compensate for a lack of a specific substance without further damaging the body by bringing in foreign chemicals or material.

Genes are the genetic units of heredity that determine what an organism will be and specific traits that an organism will possess (Becker *et al.* 2000: 10). Aside from deciding whether a plant, animal, or even an amoeba will develop, genes also influence the outward and inward workings of each lifeform. Hair color and eye color are obvious traits can be observed from parent to offspring, but other more subtle traits are controlled through genetics as well. Genes cause many debilitating diseases as well, and the ability to eliminate or repair these genes would increase the longevity as well as the quality of life for many people. The method to correct or repair faulty genes currently under scrutiny is gene therapy.

Gene therapy is a new scientific approach to treat, cure, and ultimately prevent disease by modifying a person’s genetic material (“Biotechnology in Perspective” 1990). Currently the research is focusing on delivering healthy copies of a specific gene to the cells to enable them to function for themselves, in essence allowing the body to heal itself, without the aide of heavy drugs (Wilson 1999). If proved successful, gene therapy could eliminate the horrible side effects and possibly premature death of so many who suffer from hereditary diseases.

Most individuals do not suffer from a genetic disease because they carry two copies of every gene and, in most cases, one normal, functioning gene is usually dominant enough to avoid or mask all side effects of a disease. However, problems arise when the disease is caused by a dominant mutation or occurs on the sex chromosome in males. Since males only possess one X sex chromosome, many more genetic diseases occur in males since they are linked to the sex chromosome (Campbell *et al.* 1999: 312).

### Techniques

Scientists are exploring two types of gene therapy. The first and least controversial is the notion of somatic gene therapy. Under these circumstances, an individual’s genome is altered, but the changes will not be passed to the individuals’ offspring since the germ cells are not modified. The second type of gene therapy is called germline gene therapy and it involves altering a parent’s sperm or eggs so that the changes can be passed to the offspring (Darragh 2002: 5). While somatic gene therapy is being investigated thoroughly, many object to germline gene therapy. To date the government has not approved testing of germline gene therapy. Due to such widespread controversy, this paper will focus entirely on somatic gene therapy.

Currently two different methods of conducting somatic gene therapy exist: *ex vivo* and *in vivo*. Both strategies require the use of a vector to deliver the newly modified genes into the body. Scientists are using deactivated retroviruses as the vectors of choice because of their ability to infect a variety of different cells. Some viruses even infect specific types of cells which would allow more control over where the new genes will be delivered. In addition, since the vectors are retroviruses, they have the ability to infiltrate a person’s genome and recombine with the host DNA. *Ex vivo* methods are simpler with regard to the transfer of vector and the expression of genes, but it requires surgery to remove and then replace the desired cells (Darragh 2002: 2). On the other hand, *in vivo* delivery involves targeting the desired organ, but requires minimal manipulation of the patient.

### *History of Attempts*

The first approved gene therapy procedure was performed on September 14, 1990 on Ashanti DeSilva, a four-year old child who suffered from severe combined immune deficiency, a rare genetic disease that caused an unhealthy immune system. Ashanti was vulnerable to every passing germ (Walters 1999). During the procedure, doctors removed white blood cells from her body, grew the cells in the lab, inserted the missing gene, and then infused the genetically modified cells back into her body. Ashanti was allowed to attend school for she no longer caught reoccurring colds. While this procedure was not permanent and has to be repeated every few months, it has been a huge step in giving Ashanti a normal life (Thompson 2000).

The success in Ashanti's case and other similar cases led scientists to explore gene therapy in the treatment of other genetic diseases. One case, and seemingly the biggest obstacle for gene therapy to overcome, was the treatment and subsequent death of Jesse Gelsinger. Jesse suffered from ornithine transcarbamylase (OTC), a metabolic disease that was controlled with a low protein diet and 32 various pills a day. Jesse was a rare case in a field where the diseases themselves are rare. The genetic mutation Jesse carried was not inherited, but occurred spontaneously in the womb during his development (Stolberg 1999). The treatment he received was an infusion of corrective genes, encased in an adenovirus, a weakened cold virus, which was used as the vector. After the genes were delivered, Jesse began to show strange side effects and in a few days, he was dead. September 17, 1999, the day Jesse died, marked the first death directly related to gene therapy and brought a halt to all gene therapy research (Thompson 2000).

Jesse was one of nineteen individuals who participated in the study attempting to treat, if not cure, OTC and to date no one has been able to discover what went disastrously wrong in his case. None of the other participants showed anywhere near the severity of symptoms that Jesse suffered when they received the therapy and his death remains a mystery. Some theories for the unusual death focus around the fact that his disease was spontaneously caused by random mutation, unlike the rest of the patients in the study, and the fact that he received the highest concentrated dose available (Stolberg 1999).

Due to Jesse's death, the NIH and FDA have increased regulations necessary to conform to in order for gene therapy trials to continue. It also brought about major set backs attributable to the widespread opposition of the public. To date, gene therapy research has faced major obstacles in the issues of human testing because of the publicity received from Jesse's death (Stolberg 1999).

### *Pros and Cons of Gene Therapy*

The strongest argument in favor of gene therapy is that it could eventually be used to treat desperately ill patients who have no hope of other treatment and ultimately prevent the onset of horrible illnesses (Darragh 2002: 24). The majority of candidates for gene therapy are those for whom conventional treatment and drugs have failed. Gene therapy is their last hope for a possibility of living a normal life and

hopefully a full recovery. Gene therapy also offers the perfect solution because *ex vivo* methods allow the patient's own cells to be altered and then reinserted. This would prevent major immune responses that can result in organ rejection or the body attacking transplanted bone marrow (Darragh 2002: 5). Since the donor would be the patient, all fears of rejection would be obsolete and the cells would be completely compatible. Now doctors would only have to worry about the cells performing their intended task, not agonizing over whether the cells or tissue will be rejected, possibly causing the patient more harm than good.

Opponents of gene therapy do not object to fighting disease but they question where science will draw the line. How is it possible to distinguish between "good" and "bad" uses for gene therapy and what would be the outcome if this new technology were abused (Darragh 2002: 6)? The other major issue for investigation is the long-term developments. Since gene therapy is so new, there have been no studies done to observe how patients react in the long-term since there have been no patients to examine. Another issue raised has been the notion of who should receive gene therapy if and when the methods are approved and become mainstream. The biggest fear is that only the rich would be able to afford the treatment and "the distribution of desirable biological traits among different socioeconomic and ethnic groups [will] become badly skewed" (Walters 1999). The final and possibly most convincing argument is the notion that many of the major candidates for gene therapy are children who are too young to fully comprehend and give informed consent for procedures to be performed.

### *A Look to the Future*

Despite all the controversy and setbacks, the fact remains that the wave of the future for medicine is gene therapy. Once perfected, altering or repairing an individual's genes will be the ultimate cure to the genetic diseases that have wreaked havoc on the portion of the population unlucky enough to receive mutated genes from their parents. Even if the procedure would need to be repeated, the cost for treating these individuals would decrease because they would no longer need to take regimes of pills which can cost up to five dollars a pill and can number as many as 40 pills a day. The freedom gene therapy offers would allow these patients a chance to live a normal life, not hindered by suffering or constantly needing to watch the clock to know when to take the next pill.

The other major factor is the notion that the patient's own cells can be altered and re-entered so that foreign tissue does not need to be inserted into the body. Normally genetic diseases affect one specific organ or the immune system. If transfusions or transplants are performed, the biggest fear lies in the possibility of rejection of the organ or an inflammatory immune response. These responses can cause even worse side effects than the original disease and can even lead to death of the patient. Normally when transplants are performed, the patient's immune system is weakened to the point of non-existence through drugs and then the patient runs the risk of infection from any random bacteria or virus the body can usually fight off on its own.

If gene therapy trials continue, eventually the mystery will be solved and gene therapy will become yet another tool physician have at their disposal to treat patients. The question of who receives gene therapy has arisen. The answer is simple. Everyone will be able to take advantage of the treatment. The cost for performing the therapy will be much less than the cost of all the prescription drugs and other procedures necessary. Currently the government pays for these treatments in cases where the individual themselves cannot afford the procedure, therefore there is no question as to who will receive the benefit of gene therapy.

The truth of the matter is that society cannot afford to have gene therapy be available and allow thousands of people to suffer from genetic diseases. Science will soon be able to relieve the torment that these patients face on a daily basis. How can the public claim that this is not worthy of at

least investigation? If the possibility exists to cure these people eliminate these debilitating and deadly diseases, it is our duty as a human race to at least try to alleviate the pain they suffer from.

While many may fear the implications and possibilities that gene therapy offers, the same can be said of any new idea ever birthed. People considered the notion of a vaccine preposterous and the idea that the earth was round caused the public to view people as heretics. The fact remains that many fight as well as fear any change that will alter the standard way of looking at the world, and gene therapy would alter the field of medicine irreversibly. It would eliminate the pain and suffering for patients as well as their families and could possibly be the biggest advancement in the medical field since penicillin.

## Ethics of Using Race, Ethnicity, and other Phenotypic Classifications in Biomedical Research

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*Key Words:* biomedicine, race, ethnicity, sex, gender, anthropology

### *Personal Epistemology*

I write as one whose myth for why we are here is a very simple one: that we are here because we have parents who were able to find each other, reproduce, and, in most cases, care for us. They also, in most cases, cared for each other and, living as social animals, found support from other members of their society. For me, it is from these necessities that flow all systems of ethics, that part of our understanding that can be made conscious in stories, depictions in other artistic media, and ceremonies.

I write as an anthropologist, as a member of a tragically conflicted group that celebrates the variety of human experiences possible with the many different systems that assign meaning to lives. Yet we also know that it is seldom that a culture will survive while respecting competing belief systems.

I write as a sometime forensic anthropologist who is asked by law enforcement carriers of human remains to assign the “race” of the person who in life would have benefited or suffered from a classification only sometimes and slightly registered in her bones.

I write as a man, a male whose potential reproductive period is long and whose investment in any particular offspring might be slight, possessing a nearly inexhaustible supply of sex cells. With these characteristics come all of the flaws in men so visible to women with precisely the opposite reproductive characteristics.

I write as a “white” in a society where skin color makes a

difference. White is a term that covers many ancestries; my first cousin’s parental grandparents, which make the writer an inbred, spoke German and my maternal grandparents contributed genes brought from the British Isles and then mixed with those of the original explorers of the New World.

I write as a moderately published scientist and barely published poet. Science as an epistemology shares for me one unusual distinction with the successful world religions in that it is intended to be universally applicable.

I write as a materialist, so for me, although the image of my wife is quite real even though she might not be near, there are no images of the wonderful and horrible and always interesting supernatural beings that most people keep in their head. I feel awe in an old-growth forest but do not really sense the supernatural forces that exist in an animated landscape.

I write as a teacher who nearly always failed in interesting his students in ethical questions that almost always arise in anthropological fieldwork. Such as the one I write about here.

But mostly I write as one invested in this culture, these readers, as one who may have some insights to share as to what arguments can and cannot be made about the use of race, sex, or age-group as a classificatory concept in biomedical research. Biomedical research is only one dimension of our society, of course. But in classifying participants in research by race, its practitioners behave like

we forensic scientists who classify bones by race, or we keepers of “pure” bred animals who assign personality characteristics to animals primarily maintained as “races” by selective breeding and exclusion for phenotypic characters such as color of fur. That is, biomedical research uses race in order, one hopes, to conduct better research at lower cost – research that might help people. However, what lessons are we to learn from this research? Are races so real that they even affect our health?

Consider the case of coronary heart disease (CHD). Is it possible that we will need both a biological theory of race as well as a social one in order to successfully evaluate the relations that race has with health? I had to face that question as a methodologist for a longitudinal project that attempted to learn the health beliefs of residents in central, largely rural Mississippi. Race, sex, and age-group are commonly investigated as risk factors in biomedical research. We wondered if we would be able to learn more by viewing race, sex, and age group as socially constructed by ethnic group, gender, and presented age. We were as interested in symptoms, suffering, and illness as we were in signs, symptoms, and disease. In the end, our study found that race/ethnicity was most critical in our explanatory model, that the particular interviewer was next most important, that the order of administration may have been important, that sex/gender had some influence, and that age-group was relatively insignificant. This essay is about the process and meaning of this kind of work.

#### Terms

The choice of the term sex, or gender, is like race when contrasted with ethnicity: it is conditioned by the connotation implied. With respect to health, we might be referring to, say, hormonal differences between the sexes that influence health, as in the case of osteoporosis. Or we might instead be referring to a social effect, perhaps lower rates of cigarette smoking by women than men. Social factors such as degree of social capital (Rose 2000) or social support, which can vary by gender (Linden *et al.* 1993), may be important. It seems possible that cultural models might vary between males and females within an ethnic group, or at least this is a currently popular belief. Age is another variable in which an individual’s biology and social standing both change with time, and the change may vary with ethnicity. In China I am much respected for my baldness, because age confers respect and Chinese do not bald until later years than did I.

All of these factors – race and ethnicity, sex and gender, and biological and social age – may interact in their effect on health, especially in access to health care. Social and economic standing are, in a large part, consequences of ethnicity, gender, and age and may be implicated in CHD.

In 1979, a few years before this study began, the average white per capita income in the rural, central Mississippi area was \$5,839 while for blacks it was \$2,468; about one in eight white and over half of black people lived at or below poverty levels (Shimkin *et al.* 1984). Economic factors can influence mood, and emotional affect can affect blood pressure in recurring daily life (Kamarck *et al.* 1998). These

effects may exhibit different patterns among blacks and whites. Depression, even mild depression, is more predictive of stroke than 13 other risk factors (Jonas and Mussolinio 2002).

It is possible that there are genetic factors predisposing depression that may vary in frequency among races. This is theoretically possible because races are reproductively somewhat isolated from each other and may have different frequencies of hypothetical depression-predisposing genes. Anyone can imagine that the depression experienced by members of an ethnic group, regardless of genotype, who see little hope for economic and social success could be an underlying risk factor. Mental stress, which can cause depression, can have a strong effect on CHD (Waldstein *et al.* 1996); stress is always expected for the victims of racial prejudice, and stress is known to influence cardiovascular responses since it is one of the more potent of causes of elevated blood pressure (James and Brown 1997).

However, social and economic levels have weaker associations with hypertension than with race in the west (Pickering 1999) where race also tends to determine ethnicity. Race, in the case of blacks, influences ethnicity, or one might also say caste, as well as class.

Health inequalities occur naturally in the U.S. as a consequence of racism (Dressler 1991), and stress from slights, or perceived slights, as well as more diffuse effect of prejudice (Dressler *et al.* 1999) are a factor in CHD. In urban China, to take a more ethnically and racially *Homogeneous* example, socioeconomic standing (SES) is more often associated with CHD in women (Yu *et al.* 2000). Even within SES categories and within a society, failure to meet cultural expectations, lifestyle incongruity, might predispose a member to hypertension from the resulting stress (Chin-Hong and McGarvey 1996; Dressler 1991; Dressler and Bindon 2000). But what is the relation of the biology of race and the biology of disease with the culture of ethnicity and the culturally mediated suffering from illness?

#### *Social Constructions of Identity Based on Phenotypic Characters*

In the United States race is socially constructed, and the construction is based largely phenotypic Characters of which skin color is the most important distinguishing feature of blacks and whites. Social scientists sometimes overlook the phenotypic definition of race and the reproductive isolation that has resulted from such a semi-biological classificatory system. Marriages tend to be assortative with respect to skin color, which maintains partial reproductive isolation among different skin color groups. It can be noted that the importance of the tendency to select mates like one-self was emphasized by Darwin (1871). Inheritance can be measured as the proportion of variance of a quantitative trait that is explained by heredity (Falconer 1981). The effect of assortative mating on a polygenic trait such as skin color is to increase the strength of inheritance of the trait (Cavalli-Sforza and Bodmer 1971: 543). However, traits not salient for assortative mating should not be preserved in groups defined by these salient traits, unless they should be, as we imagine for the behavioral traits of our “races” of dogs and

horses, perfectly linked with the underlying genes. Even for traits with an effect from a major gene, unless tightly linked with a “racially” defining trait by location on a chromosome, will not remain associated with skin color over generations of mating. Biomedical scientists who intend to use race as a proxy for population differences sometimes fail to understand this important consequence of quantitative genetics. Social scientists have labored under even greater misapprehensions.

A trait with some heritability, such as one with a score on the Stanford-Binet test, has incorrectly been associated with genetic differences among races (Hernstein and Murray 1991). The heritability of what is perhaps the most investigated questionnaire, the Stanford-Binet Intelligence test, measured in a broad sense (Falconer 1981), is high in some populations, often higher than the heritability of most diseases. Differences among individuals in a population have a substantial genetic component. Differences in intelligence test performance are also noted among ethnic groups. Blacks, on average, are poorer performers than whites. Waldman *et al.* (1994) found no association between the number of marker genes, especially those higher in frequency in Africans than in Europeans, among black test takers, so we may conclude that the average differences in test scores among populations do not have a significant genetic component even though differences among individuals in a population may.

Other data are also available that suggest the differences among races in IQ scores may vary much more by experience of the test taker than by her genes. When mean IQ scores are de-standardized by year of administration so that absolute values can be compared across years, we find that average IQ scores have risen dramatically over the last 75 years in a variety of countries where such records have been kept. The increase in average score is almost double the magnitude of differences observed today among ethnic groups. IQ tests are given in childhood to children whose childhood environments vary by ethnicity. If one postulates that the environment can become matched to IQ score, a likely outcome in a semi-meritocracy such as the U.S. where “intelligence” is related to access to wealth, the environment can produce a very strong effect even if the heritability of IQ within a population were in fact high (Dickens and Flynn 2001).

In another study, a meta analysis of indicators such as intelligence tests, personality, and social factors found that even the few percent of the variation that might seem attributable to racial differences disappeared when adjusted for socioeconomic factors (Gorey and Cryns 1995). From Dickens and Flynn we can see that even a heritable aspect of race can still carry very strong environmental effects on a traits such as, for example, response to a questionnaire, or blood pressure.

Despite the problems in interpretation of black/white differences, some of which have been suggested above, we proceed with that difference conceptualized primarily as cultural. We may be justified in this specific case because previous training of community health facilitators as cultural-brokers was successful in this rural Mississippi

population (Storer and Frate 1990; Whitehead *et al.* 1984). Success must have required an understanding of ethnic differences of belief systems about illness. Certainly the degree of overlap of the social worlds (Tovey and Adams 2001) of the subgroups with those of the caregiver needs to be understood in order for culturally sensitive interventions to be successful (Storer and Frate 1990).

Gender and age group are also commonly constructed by the viewer from external observable phenotypic and cultural traits such as hair treatment, clothing, surface treatment of skin, and so forth. Judgment of gender, for example, can be influenced by individual response to hormones, external genitalia, or chromosomes. Age is judged by actors and observers by observable effects of aging and possibly influenced by aids to deception of actual chronological age. My wife is a year older than me but appears 10 years younger. I can carry heavy loads in the Andes further and quicker than some of my students 30 years younger than me. But can we really cheat the effects of race, sex, and age in fighting disease? Is there not some underlying genetic basis of these categories regardless of how much deception, or self deception, that we practice?

#### *Biological Constructions of Identity Based on Genotypic Characters*

To the extent that dark skin indexes frequencies of genes, and if there are major or polygenetic genes that influence coronary heart disease risk, then race would predict cardiovascular disease incidence and be a legitimate risk factor for any physician to consider for her patient. Rates for some diseases differ between 5 and 100 fold among “races,” which tempts one to assume that these differences are due to genetic factors. However, the magnitude of these differences may also have resulted from culturally different life experiences. Evidence for this proposition is that when low-risk groups emigrate to a high-risk country, their risk will usually increase although their race remains stable (Willett 2002). Of course, the presence of differences between countries does not preclude genetic effects within a country.

#### *Interactions of Cultural and Biological Factors in Studies of CHD*

To the extent that dark skin indexes class, which itself may influence risk factors such as stress, diet, activity, smoking, and alcohol consumption, then race, as ethnicity, will predict risk. In the United States and elsewhere, race, sex, and age directly influences social and economic class, and each can lead to failure to enjoy the privileges of the class that one might otherwise be entitled. Race is an especially salient classification because of the automatic assignment of darker skinned people to a lower class. The effect is especially strong for males (Dressler 1991).

If the stereotypes of gender, possible because of observable biological differences between the sexes, effect an individual’s recognition of cardiac-related symptoms, then gender too may predict risk (Martin *et al.* 1998). Another gender-linked example is that lack of social support may be a predictor of blood pressure levels in women but not men (Linden *et al.* 1993). Further complicating the

phenotypic landscape, ethnicity, in the case of blacks and whites, may function differently by gender. For example, consider the finding that American white women in clerical jobs experience a much greater drop in blood pressure during sleep than do black women in the same job (James 1991). These findings suggest that either stress affects the two races differently or that different types of stress are presented to one group compared with the other.

When viewed as cultural constructions, ethnicity, gender, and age are components of culture. American anthropologists often take a holistic perspective in order to understand the human condition. In the case of illness, this requires taking into account language, behavior, beliefs, genes, physiology, and the forces of evolution. Thus groups, defined by culture, which are genetically semi-isolated populations, defined by reproduction, are analytically separate concepts that may be exhibited in a single group of people, at least for some generations.

#### *CHD and Culture*

Dressler and Bindon (2000) review the use of the concept of culture in the study of social factors and health. The question that arises when dealing with black and white ethnic groups is whether it is ever appropriate to treat blacks and whites as participating in the same culture with respect to illness. In a famous study of hypoglycemia in the Andes Bolton (1984) argues that the inhabitants of a single region in southern Perú and Bolivia, even though they spoke different languages, should be considered as a single culture, the Quolla (but see Lewellen 1981 for a counter view, one which our experience there would support).

Should blacks and whites, the participants in the Mississippi study that I have participated in, who speak dialects of the same language, who are poor, and who live in the same region, be treated as a single group or as two? The approach we adopted was to treat them provisionally as a single group with respect to beliefs about health and then evaluate underlying dimensions of beliefs by ethnic groups on a dimension-by-dimension case. We used the same design for the factors of sex and age, withholding judgment as to any systematic differences in health beliefs between genders or among age-groups until the evidence is tabulated.

However, if there are systematic differences in understanding of illness by sub-culture or ethnic group, further questions will be suggested. Will response to social stress or lack of social support vary among groups? Would these differences, if they exist, be more apparent in belief systems than in social behavior or in tests? We selected questions about beliefs for this study although we know that both beliefs and behavior must be studied for a full understanding of a culture (Guillet *et al.* 1995). We created a special questionnaire for this study.

But why not use standard psychological tests to understand health? One reason is that traditional psychometric indexes do not always relate significantly to CHD (Coelho *et al.* 2000). We suspect that this is so because the terms are not understood by individuals or perhaps because the questions asked are not reasonable within the conceptual framework of the individual, especially when the

culture of the designer of the test differs from the respondent. For these reasons, our instrument addressed these problems by eliciting culturally salient terms and questions that might be reasonably asked about them. We hoped to represent the health beliefs shared most widely among the target populations in our questionnaire.

We know that variability in degree of sharedness of the beliefs of the general cultural model may be associated with blood pressure levels (Chin-Hong *et al.* 1996; Dressler and Bindon 2000: 258). In fact, in the central rural Mississippi area, social marginality, which is a lack of sharedness, was identified as a risk factor for CHD (Shimkin *et al.* 1984).

#### *What Are We Able to Conclude from this Study?*

Briefly, we found that interpretable dimensions of a model of health beliefs emerged even when all participants were included. Race, interviewer, and order of administration were the major factors in which individuals differed across the dimensions. Risk of CHD was found to be associated with significant interactions by race and other factors. We concluded that knowledge of the breadth of variation in health beliefs by race would be important in educational interventions. We were surprised to find how reflexive were the limited responses permitted to our questionnaire by interviewer and across repeated administrations. The social effect of the oral interviews was greater than anticipated.

What can we say about the use of race, sex, and age-groups in biomedical research? First, we cannot say that simply because these categories are labile there are no differences in incidence of diseases across these categories. Some disease incidences are 500 times greater in one race than another. The question is, when are these differences due to genetic variation, such as in Thalassemia for Mediterranean populations, or the less severe blood disease Sick-cell anemia among western African populations and their descendents? When are these different incidences due to the use of phenotypic characters to classify one into an ethnic group, itself the salient factor? We can't be sure.

Social scientists sometimes want to believe, as part of their mythology, that there are no important differences among the races. Biological scientists, with another mythology that supports their worth, sometimes want to believe to the contrary. Racists, of course, have still another mythology to support. What effect does biomedical research or forensic identification, or the keeping of "races" of animals, have on our understanding of the obvious differences among ethnic groups defined by phenotypic characters, and the meaning of such differences?

First, classification by races on the basis of observable phenotypic characters exists and has a powerful effect on the lives of the classified. A question is to what extent an individual may avoid some of the deleterious effects of being a member of the race associated with the lower class, such as blacks in the US, untouchables in India, or the Hutu in central Africa? One has to hope that education may, in the very long run, ameliorate the connection with class. In the short run, interventions of the type that were made in central, rural Mississippi, can be effective.

But couldn't this research have been done without recording race, by simply recording SES variables? That depends on whether the reader believes that to grow up black and poor is the same as to grow up white and poor. In our samples, like most, the poor whites were less poor than the poor blacks; thus, we were unable to investigate this question. As anthropologists, we suspect that many beliefs are relatively common among the poor and that many beliefs

are relatively common among blacks or whites, due to their different lived experiences. So, in the end, was I ethically compromised to participate in a study that was phrased by racial differences? Yes. Did our work do anything to alleviate these differences where they impact health? It is too soon to tell, the results are just now being published. What did I learn? I now know how very difficult this kind of research is, but, Lord help me, how very fascinating.



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