

Human technology, evolution and sickle cell disease

[Science](#), [Biology](#)



Claim

Human technology has caused evolution of the human species to stall

Research question

To what extent has advancements in medicine helped preserve negative genetic mutations in the human gene pool?

Rationale

In “ On the Origin of Species”, Darwin proposed that changes occur in the characteristic of a species over several generations. Because resources are limited in nature, organisms with heritable traits that favors survival are chosen to reproduce and pass on their genes to the next generation. Mutations, or random changes that occur in the DNA sequence, can be neutral, beneficial, or harmful to the organism’s survival. While advantageous mutations are selected “ for” to reproduce, disadvantageous mutations are often selected “ against”, decreasing the mutated organism’s chances of surviving and reproducing. However, with the advancements in medical technology and the discovery of new treatments for genetic diseases, a mutated individual’s survival chances and life expectancy has increased greatly, providing them the opportunity to reproduce and pass on the mutated gene to their offsprings. Therefore, a negative genetic mutation that otherwise may have been expunged by natural selection now has the opportunity to be inherited by new generations and remain in the human gene pool.

Background Research

Sickle cell disease (SCD), also known as sickle cell anemia, is a group of genetic disorders that causes abnormal hemoglobins in red blood cells. It is passed from generation to generation in an autosomal recessive pattern, which means both copies of the gene in each cell are mutated. Severity may vary from person to person, and signs and symptoms of the disease begin in early childhood.

Bacterial infections, most notably *Streptococcus pneumoniae*, has been noted as a major cause of death of SCD. As such, a Penicillin Prophylaxis in Sickle Cell Disease (PROPS) study was launched in 1983 to determine whether daily administrations of penicillin would reduce the occurrence of pneumococcus in patients under the age of 3. In the first phase of PROPS in 1987, a multi-center randomized control trial was initiated with treatment of penicillin and placebo groups. An approximately 84% reduction in pneumococcal septicemia was found in the treatment group with penicillin.

The trial was terminated 8 months early due to the definitive results, and it was concluded that children should be screened at birth for SCD and provided prophylactic penicillin at 4 months of age to avoid infection by pneumonia. In comparison to a study in 1975 which found an overall mortality rate of 7.3% in patients less than age 23, the overall mortality rate of 2.6% in children less than age 20 in 1987 indicates gains in survival, largely due to the implementation of penicillin prophylaxis. Comparatively, in February of 2000, the development of the 7-valent pneumococcal vaccination was followed by a study which demonstrated that vaccination of

pediatric patients with the 7-valent vaccine beginning at 2 months of age, followed by vaccination with the 23-conjugate vaccine at 2 years of age, resulted in a safe and elevated antibody concentration of 7-valent pneumococcal serotypes. In March of 2008, a study further validated the use of 7-valent vaccine in children under age 10 with SCD, observing a reduction of 68% in pneumococcal infection post the licensure of the 7-valent vaccine in 2000. Therefore, it can be concluded that the institution of penicillin prophylaxis and pneumococcal vaccination were both critical in the improvement of childhood survival rates.

Analysis and Interpretation

In 2004, the National Institutes of Health-sponsored Cooperative study of sickle cell disease estimated that the median survival age was 42 years for men, and 48 years for women. The median age of death does not seem to have improved since the publishing of *Mortality In Sickle Cell Disease—Life Expectancy and Risk Factors for Early Death* in 1994. However, an increase in the number of patients over the age of 45 can be observed. SCD-related mortality during childhood contributed significantly to this shortened life expectancy, although improvements in the medical care of children have since increased their survival rates. Measures and treatments include early diagnosis by newborn screening, parental education, and the previously mentioned prophylactic penicillin to prevent fatal pneumococcal sepsis, as well as more recent advancements such as hydroxyurea, stem cell transplantation and the expansion of chronic transfusion programs.

These medical advances continuously improve survival, reduce disease-related morbidity and improve quality of life for SCD patients, particularly during childhood when patients are more at risk of pneumococcal diseases. However, treatment can only manage the associated signs and symptoms of SCD, not the underlying mutation in the genes. Both men and women now survive well into their reproductive years, and many choose to reproduce despite the high risk of transmission. It is estimated that SCD occurs among 1 of every 365 African-American births, and 1 in 13 African-American babies are born with the sickle cell trait, which makes SCD one of the most common genetic disorders.

Based on the high rate of mortality in children with SCD without the availability of modern treatment, it can be assumed that natural selection would have acted on sickle cell disease by eradicating the deleterious alleles, which would cause the number of SCD patients to decrease over time. Similar assumption can be made for other genetically inherited diseases, such as cystic fibrosis and Tay-Sachs disease. In most patients afflicted with genetic conditions, reproduction is now considered a personal choice that may be a significant factor in the future's scientifically advanced society. Therefore, advancements in medical care has slowed, if not completely stalled, the process of natural selection, and consequently, human evolution.

Evaluation

This investigation is based upon the statistics and data from scientific journals, which can be considered accurate and reliable as most have been reviewed by subject-specialists before publication. As supported by the data,

human technology and the discovery of new treatments for sickle cell disease has greatly decreased mortality rate among children, allowing more SCD patients to survive to adulthood and reproduce. However, the statistics used in this investigation is limited to African-American patients in the US. It does not account for patients in Sub-Saharan Africa and South Asia where SCD births are more common because of the resistance to malaria that the prevalence of the sickle cell trait provides. SCD-related mortality in Sub-Saharan Africa and South Asia may also be higher due to less available medical care and treatment.

Cases of malaria are rare in the US, and therefore the sickle cell gene is considered deleterious as the risk of infection is low. On the other hand, the sickle cell trait may be beneficial to an individual's survival in countries such as Nigeria and Uganda where the malaria disease is more common. The investigation could also be improved by comparing the number of children born with SCD from the 1970s to the 2010s and analyzing whether occurrence of the sickle cell allele has increased since the discovery of newer treatments. An increase in children born with SCD would further support the claim " human technology has the evolution of the human species to stall".

Conclusion

Medical advancements and new treatments for genetically inherited conditions has significantly increased survival rates among affected individuals, particularly young children who are more likely to succumb to infectious diseases. Therefore, with the increase of affected individuals

reaching reproductive age, the process of natural selection and adaptation over generations is hindered, and evolution is stalled.

References

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