Devin Carree

10/7/19

IBIO 445

H. Sapiens Potential for Evolution through microevolutions associated with Autism Spectrum Disorder

Introduction

In 1859, Charles Darwin published his book, "The Origin of Species" on the theory of evolution (Darwin, 1859). During a time of low evidence and high skepticism, Darwin used his scientific deductive reasoning to postulate the idea that somehow every organism that ever existed evolved from one common organism or ancestor. There were as many skeptics back then as there are now, but with advanced scientific technology and more scientific knowledge many of these ideas have not been challenged or have been improved upon. Some skeptics today cite the non-existence of our predecessors as a significant lack of information to disprove the theory of evolution, while some may cite the non-existence of our future evolved forms. While current researchers are searching for more evidence and information into Neanderthals and our ancestors, this paper suggests that it is possible that an advantageous gene, set of genes or neuro-anatomic phenotype may arise in the human population through genotypic mutations often phenotypically characterized as Autism Spectrum Disorder.

Autism Spectrum Disorder (ASD) is a common "disorder" that affects 1 in 68 people in America and extends globally (Bahado-singh, 2019). Autism is a disorder that has yet to be fully defined as it presents itself in many ways. It is currently suggested that ASD is a neuro-developmental disorder characterized by impaired communication difficulties, social interactions and/or repetitive or restricted patterns of behaviors, interests or activities (Treffert, 2009). This disease is extremely heterogenous and presents itself as many different phenotypes. Autistic disorder, Rhett syndrome, Asperger syndrome and childhood disintegrative disorder, once all recognized as different pathologies, are now collectively known as ASD. The comorbidity of ASD is also very high. Autism is often present alongside many other disorders such as attention-hyperactivity deficit disorder (ADHD), bipolar disorder, anxiety disorders, intellectual disability, schizoid personality disorder, obsessive—compulsive personality disorder, Tourette's Syndrome, etc. This makes Autism extremely hard to define without the inclusion of symptoms of the other ailments. Although many patients subject to ASD suffer from these symptoms and issues, as many as 1 in 10 people who suffer from ASD also are affected with a gain of functionality

disorder known as "Savant Syndrome". Savant Syndrome is a rare condition in which a person with a serious mental disability such as ASD has enhanced skill or an "island of genius" in fervent contrast of the actual disease (Hurst, 2019). It is estimated that 50 percent of people with Savant Syndrome also presents with ASD. The gain of function aspect of Savant syndrome is typically centered around memory and can be narrowed to five different areas: music, art, calendar calculating, mathematics and mechanical or spatial skills. These skills include, perfect pitch, the ability to hear any musical note one time and reproduce it in many/all different instruments, the ability to calculate the amount of hours in 40 years accounting for leap years without a calculator, the ability to remember and recite a very long list of words, verbatim, or the ability to calculate distance with a high level of accuracy using only vision (Treffert, 2019). This ability could be used as an evolutionary advantage in a future society.

In 1973, Russian geneticist and evolutionary biologist Theodosius Dobzhansky said, "Nothing in biology makes sense except in the light of evolution." (Dobzhansky, 1973) Now over 100 years later we can not only use Darwin's postulate of evolution to understand and create things helpful or essential for survival today, such as vaccines and anti-bacterials, but also look deep into the future for "sci-fi" like ideas or hypothesizes about life in the future. An advantageous set of genes or neuroanatomy may be incorporated into the gene pool of human beings, allowing for a speciation event to occur in the human species. The universe is estimated to be approximately 13.8 billion years old. The oldest hypothesized eukaryotic life on earth is hypothesized to be approximately 1.5 billion years old. Human and primate lineages have been hypothesized to have diverged 6 million years ago. The final lineage of prehistoric "human", before modern day humans, went extinct 1.9 - 0.05 million years ago. Leaving the Homo Sapiens, modern day humans, as the last extant species of human. This speciation event is estimated to have occurred 350,000 years ago (Schelebusch, 2017). This means that the modern-day human has been around for over 350,000 years and have been subject to over 348,000 years of mutations before the common era and more than 2000 years of mutations in the common era. There are five major elements tied to the theory of evolution (Darwin, 1859), three of which are relevant 1) microevolution- the change in genes in the genome over time, such as the change between a typical human being and the genetics of an autistic human being, 2) speciation- the process by which species/lineages split and diverge over time and 3) macroevolution – the dramatic shift in phenotypic forms of life over long periods of time, often creating novel forms of life. A defining feature in the differentiation of the human lineage from other primate species is "Habitual bipedalism" (Pontzner, 2017). Laboratory studies show that the way modern humans walk is much more energetically favorable than chimpanzees, covering nearly twice as much distance using the same number of calories. It is hypothesized that this provides an evolutionary advantage, allowing early hominids to use less energy for increased reproduction, larger brains and other metabolically costly traits. This suggests that selection for endurance capabilities played a key role in shaping human lineages. The memory ability of a human being with savant syndrome like abilities may play a similar role in the evolution of these same human lineages.

Body

Evolution has been occurring since the beginning of life itself, for more than 4 billion years. Darwin postulated that every species that exists today evolved from one single celled organism (Darwin, 1859). If this is true, it can be reasonably assumed that in the human population today, evolution is still occurring. Evolution has no effect on individuals. Evolution influences populations by introducing different genes into populations through mutations causing differentiation and differences within the same species, this is microevolution and can be seen in autistic patients. Genetic disposition only accounts for 10% of autism cases, and have no single genetic factor (Sealy, 2016), but it is estimated to have over 90% heritability (Lichtenstein, 2010), which suggests a strong genetic component. An example of these microevolutions includes de novo duplications of chromosome 7q11.23 which is associated with ASD significantly (Sanders, 2011). Five additional regions with rare recurrent de novo copy number variation mutations have also been identified as being associated with ASD. To be able to witness a difference in the population of human beings, or macroevolution, the human gene pool would have to have a significant number of humans with the correct genetic mutation while also having a 'stable' enough genome to function in society. Some studies suggest that for the past 2 decades the prevalence of ASD has steadily increased. In the U.S. from 2000-2010 ASD increased from 0.67% to 1.47% in the U.S. before reaching its first plateau of 1.46% in 2012. It then continued to increase in 2014-2016 to 2.46% with no significant increase within these years (Xu, 2018). A population of human beings associated with ASD could eventually lead to a speciation event in the entire human population. There may be people in society today that will be associated with the speciation of human beings that carry the advantageous genes but do not present with ASD. Although ASD is the disorder that carries the advantageous set of genes or neuroanatomy, the evolved form of the human being is not a human being with autism, they are the link between modern day human beings and higher functioning future human beings.

In evolution there are organisms known as transitional forms. Evolutionary biologists define transitional forms as the intermediate species with a mixture of features between both life forms. An example of transitional forms would be Pacific leaping blenny fish *(Alticus arnoldorum)*. (Madelaine, 2004) They are a great example because they may help connect the evolution of life in water to

Perciformes. Perciformes are an order of fish that have evolved many adaptations to survive out of water. One of the most common adaptions, lung like organs, could help explain the evolution of lungs in terrestrial animals. Many people concerned with the evolution of human beings are generally concerned with our concrete ancestors such as *H. erectus or H. neanderthalensis* but there were also transitional forms in humans. *H. luzonensis* or *H. heidelbergensis* which each had genotypic and phenotypic differences from their ancestors, such as intra-cranial volume expansion. This may have contributed in the elaboration of stone tool technology (Schlebusch, 2017), may have also contributed to the creation of *H. sapiens*. The subspecies of human beings with ASD would play a similar role in the speciation of *H. sapiens*.

It is possible there are human beings in the population today with an advantageous set of genes or neuroanatomy associated with ASD, without displaying symptoms of the actual disorder itself. If this is true, over time the population would begin to see a significant phenotypic increase in memory abilities, calculation abilities and/or evolved senses (sight, smell, touch, hearing, taste), and this gain of function ability would be located within the genotypic mutations involved in the phenotypic diagnosis of ASD, but these individuals would not be diagnosed with ASD. Genetic disposition accounts for 10% of autism cases, but it is estimated to have over 90% heritability. Because DNA (genes) are the heritable source of information between generations, this may be confusing. A single gene may not be the cause of the development of ASD, instead this implicates genome by environment interactions, or epigenetic interactions. Epigenetics is defined as changes in genetic expression and gene function not due to mutations. Current evidence suggest that this could play a significant role in the onset of ASD (Bahadosingh, 2019). This could also help decipher the many comorbidities that are present alongside ASD. One or many similar chromosomal mutations that allows for the advantageous gene, set of genes or neuroanatomic phenotype characteristic of an "evolved" human being may also allow for phenotypes such as attention-hyperactivity deficit disorder, bipolar disorder, anxiety disorders, intellectually disability, schizoid personality disorder, obsessive—compulsive personality disorder, Tourette's Syndrome, etc. through epigenetic mechanisms. Epigenetics is a new and very active area of research.

Conclusion

If Charles Darwin's theory of Evolution is true, then evolution has been occurring since the first life form to ever exist on earth through genetic mutations and selection. Some people don't believe in evolution, but as Theodosius Dobzhansky said, "Nothing in biology makes sense except in the light of evolution." If every form of life in the world is evolving and have been evolving since the beginning of time it would only make sense that humans too are evolving. The inability to think about the possibility that the universe was not created for human beings or our own species can make this idea hard to conceptualize. Someone who suffers from this would believe that humans are the most intelligent form of life there ever was and ever will be. In this light there is no way that humans could ever have evolved from primates and there is no way that humans can evolve into a more intelligent organism. But in the light of evolution, where fish evolve into terrestrial animals by the development of air-breathing lungs from water-breathing gills and the development of arms and legs by the evolution of muscular fins that are able to propel fish across land, the evolution of Human beings make sense. If primates began walking on two feet, conserved energy to enhance hunting ability, and evolve increased cranium space to enhance thinking ability to evolve into a human being, then it makes sense that a human being with a differentiated neuroanatomy and savant abilities may have an increased or more efficient thinking ability which may lead to the evolution of the human being.

It is possible to evolve in many ways. Such as neurologically, muscularly, immunologically etc. Scientist suggest the one of the most recent ways that humans evolved is through the microbiome because some of our ancestors, relatives and even some of the species cannot digest or metabolize lactose while others can. But because the most significant evolution event that occurred was neurologically, the next significant evolutionary event may be neurologically as well and it may be located within ASD.

Work Cited

- Bahado-singh RO, Vishweswaraiah S, Aydas B, Mishra NK, Yilmaz A. 2019. Artificial intelligence analysis of newborn leucocyte epigenomic markers for the prediction of autism. Brain Res 1724:146457.
- 2. Darwin, C. (1859). The Origin of Species. Oxford: Oxford University Press, pp.155, 158.
- Dobzhansky, T. The American Biology Teacher, Vol. 35 No. 3, 1973; (pp. 125-129) DOI: 10.2307/4444260
- 4. Hurst TP, Magiorkinis G. 2019. Editorial: The Past and the Future of Human Immunity Under Viral Evolutionary Pressure. Front Immunol 10:1–3.
- 5. Lichtenstein P, Carlström E, Råstam M, Gillberg C, Anckarsäter H. 2010. The genetics of autism spectrum disorders and related neuropsychiatric disorders in childhood. Am J Psychiatry 167:1357–1363
- 6. Madelaine, B (May 2004). Migration history of air-breathing fishes reveals Neogene atmospheric circulation patterns. Geology 32 (5): 393–396.
- 7. Matelski L, Van de Water J. 2016. Risk factors in autism: Thinking outside the brain. J Autoimmun 67:1–7.
- 8. Pontzer H. 2017. Economy and Endurance in Human Evolution. Curr Biol 27:R613-R621.

- Sanders SJ, Ercan-Sencicek AG, Hus V, et. al 2011. Multiple Recurrent De Novo CNVs, Including Duplications of the 7q11.23 Williams Syndrome Region, Are Strongly Associated with Autism. Neuron 70:863–885.
- 10. Schlebusch, C; Malmström, H; Günther, T et al. (2017). "Southern African ancient genomes estimate modern human divergence to 350,000 to 260,000 years ago". Science. 358 (6363): 652–655.
- 11. Sealey LA, Hughes BW, Sriskanda AN, Guest JR, Gibson AD, Johnson-Williams L, Pace DG, BagasraO. 2016. Environmental factors in the development of autism spectrum disorders.Environ Int 88:288–298.
- 12. Treffert DA. 2009. The savant syndrome: An extraordinary condition. A synopsis: Past, present, future. Philos Trans R Soc B Biol Sci 364:1351–1357.
- 13. Xu, Guifeng et al. "Prevalence of Autism Spectrum Disorder Among US Children and Adolescents, 2014-2016." JAMA vol. 319,1 (2018): 81-82.