

Journal of Psychology and Psychiatry Studies

Stickler Syndrome and Schizophrenia: Congenital Vs. Genetic

Robert Yousif*

¹6116 Castilla St. Santee, CA, 92071, United States of America

While reviewing the charts of a patient that was coming in, I noticed her son was diagnosed with Stickler Syndrome. Looking deeper into her family history, her daughter was blind, and the patient herself was suffering from schizophrenia. At the time I was doing my core rotations in Baltimore as a medical student and had not known what stickler syndrome was nor had I ever heard of it.

I had the opportunity to speak with her when she came in, asking questions as I normally would with any patient while obtaining the patient history. The woman was in good physical shape, appearing generally well. She told me stories about how her washing machine is recording her, and the radio is sending her messages. She followed each of these statements with 'I know it sounds crazy'. Also, an interesting side note, the patient was also hypervigilant when it came to her mammograms-her chart showing that she would get 1 every 6 months for the past 10 years. Nonetheless, her previous diagnosis was confirmed.

Schizophrenia has many varying degrees of severity and manifestations, though the cause largely remains a mystery. Although genetics surely do play a big factor, as more than 40% of monozygotic twins with schizophrenia are both affected [1], the genes known to be involved in schizophrenia each have a small effect and unknown transmission and expression [2].

Environment also plays a role in the cause of schizophrenia. Maternal stress has been associated with an increased risk of schizophrenia, possibly in association with [reelin](#) [3], While both maternal stress and infection have been demonstrated to alter fetal neurodevelopment through pro-inflammatory proteins such as IL-8 and TNF [4].

Marijuana use also has been implicated in the risk of causing schizophrenia. The most recent study being in August 2018, in which researchers from Radboud University, published in Nature Neuroscience, looked at data from more than 180,000 people as part of the study, which identified 35 different genes associated with cannabis use with the strongest associations in a gene called CADM2. The study also found a genetic overlap between cannabis use and the use of tobacco and alcohol, also a similar overlap between cannabis use and personality types that were prone to more risky behavior or were more extraverted. Although the study uncovered that people with schizophrenia are also more likely to use cannabis, the study concluded that people with a vulnerability to develop schizophrenia are at increased risk of using cannabis.

Genes that have been implicated schizophrenia include 22q11, 1q21 and 16p11 [5], while genes that have been

Article Information

Article Type: Analysis

Article Number: JPPS103

Received Date: 06 September, 2018

Accepted Date: 12 September, 2018

Published Date: 19 September, 2018

***Corresponding author:** Dr. Robert Yousif, 6116 Castilla St. Santee, CA, 92071, United States of America. Tel: 6199442186, Email: [robertyousif6\(at\)gmail.com](mailto:robertyousif6(at)gmail.com)

Citation: Yousif R (2018) Stickler Syndrome and Schizophrenia: Congenital Vs. Genetic. J Psychol Psychiatry Stud 1: 103 (21-22).

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implicated in stickler syndrome include COL2A1 in region 12q13.11-q13.2, COL11A1 gene in region 1p21, and COL11A2 gene in region 6p21.3. These genes show varying phenotypes of stickler syndrome, in which the different types have some or all of the characteristics, which are hyper extensible joints hearing loss, and severe near sightedness with associated eye problems. The typical facial features include flattened bridge of the nose and flat cheekbones. The Robin sequence (or Pierre Robin sequence), is also common in children with Stickler syndrome which includes a U-shaped cleft palate with a tongue that is too large for the space formed by the small lower jaw. The eye findings may include changes in the vitreous of the eye, and increased risk of glaucoma, retinal detachment, and cataracts. Inheritance is thought to be autosomal dominant, or due to a new mutation.

The human genome project, which was declared complete in 2003 with the objective of determining the DNA sequence of the entire euchromatic human genome. The project has helped us understand genotyping and mutations of specific types of cancer [6]. But many thought this project would give us all the answers to genetics we could ever want. But since the completion of the project it became clearer that knowing the gene sequence or location of a disease tells us little about the cause or possible treatment plans. Gene transmission and expression are a much more mysterious and important part of the formula. So how much useful information does locating the gene(s) of a particular disease actually tell us?

The causes of any disease fit into one of three categories: congenital, genetic, or environmental. But if we can locate the gene mutations that cause a certain disease, and we find there to be a statistically significant association between another disease, and these diseases share no similar gene mutations, the only logical conclusion would be to eliminate the gene location as a cause; though gene expression and transmission may still play a role. Which now leaves us with congenital or environmental. Congenital diseases are due to a defect with the complex process that begins with

conception and ends with-well, what we all are who are reading this. Given proper prenatal care, these diseases tend to be random events with no correlation in disease prevalence within certain populations. Which leaves us with environmental causes. This is the broadest of the three causes as it encompasses everything from stress, to infection, to drugs, to pollution, and everything in between. With so many possibilities, environmental causes are the hardest to pinpoint. But if we are able to find a statistically significant family correlation, are we still able to point to environmental as the cause?

Worldwide schizophrenia prevalence is 1 percent, while stickler syndrome prevalence is 0.03 percent. This makes the odds of two random people each having one of these diseases about 0.0003 percent. As a medical student now in my 4th year of clinical rotations I can only speak on the limited clinical experience I have had. But possibly we need to add a more statistical approach to the current genetic approach to determining which diseases are associated with each other or likely to have a higher prevalence in a population with a certain disease, as genetic transmission and expression and what effects each of them largely remains a mystery. Could schizophrenia be linked to stickler syndrome, or blindness? My sample size was too small to make such conclusions, but further investigation is warranted.

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