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Medical Norm of Psychiatric Misdiagnoses in Rare Disease Patients

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In medical school, students are taught this adage: “When you hear hoofbeats, think horses not zebras” (Cormier 196). This adage indicates that when doctors diagnose patients, they should default to common conditions rather than rare ones. However, this practice has multiple negative ramifications for patients with rare diseases. A rare disease patient myself, I have experienced firsthand these repercussions. For most of my childhood, I was misdiagnosed with growing pains and depression. Prior to an accurate diagnosis, due to lack of appropriate treatment, my symptoms of joint subluxations and chronic pain grew progressively worse each year. Simple tasks of daily living such as combing hair and brushing teeth became increasingly difficult. It was not until the age of seventeen that a specialist correctly diagnosed me with a rare, genetic connective tissue disorder called Ehlers-Danlos Syndrome. Upon my diagnosis, I connected with others in the rare disease community where I discovered that unfortunately, my experiences with psychiatric misdiagnoses are far from unique. The medical norm of psychiatric misdiagnoses in rare diseases patients not only ignores the possibility that future medical advancement or more exhaustive testing will reveal a biological cause but also has harmful ramifications for their physical and mental wellbeing by delaying necessary treatment whilst questioning their sanity.

This medical norm utilizes ‘the psychogenic inference’, which infers that "if there is no known physical cause for a symptom or disorder, the cause must be psychological” (Sykes 289-290). Physicians use the term medically unexplained symptoms (MUS) to classify symptoms for

which no biological cause has been found (O'Leary 7). MUS can range from mild to very severe and can include chronic fatigue, dizziness, muscle pain, digestive issues, and neurological issues (Brown 789). Medically unexplained symptoms are more prevalent than most would expect. According to a 2007 study, 10-15% of patient visits to a general practitioner in the U.S. are due to severe medically unexplained symptoms (Brown 773).

Given that only about 0.1% of the U.S. population suffer from rare diseases ("FAQs about Rare Diseases"), the vast majority of the estimated 10-15% of total US patients with MUS visiting their primary care doctor will not have a rare disease. From a global perspective, a study found that rare diseases affect approximately 3.5–5.9% of the global population, which equates to approximately 263–446 million individuals (Nguengang 165). Although this percentage is very small, when the actual number of individuals in the percentage is considered, it becomes evident just how many individuals are affected by rare diseases and thus personally encounter the norm of psychiatric misdiagnoses.

This harmful medical norm delays necessary medical treatment for rare disease patients. One such example can be found in the diagnostic journey of Chloe Atkins, a professor at the University of Calgary. Atkins suffers from myasthenia gravis, a rare autoimmune disease that disrupts communication between nerves and muscles (Atkins 20). However, for fifteen years, the medical field accused her of fabricating the muscular paralysis caused by this disease (Atkins 20). She asserts that an accurate diagnosis was the difference between spending the rest of her life in a wheelchair and living her current life as a researcher and tenured professor. (Atkins 20). Another occurrence of this norm was recorded in Japan, where a woman was misdiagnosed with a panic disorder until a doctor finally discovered that she had Graves' disease, an autoimmune disease associated with hyperthyroidism (Yasuda 2).

These two stories become more disturbing when considering the recent rise in the prevalence of uncommon diseases, resulting in an increase in the amount of people affected by this norm. The National Institutes of Health (NIH), a leading global biomedical research agency, performed a study specifically geared toward calculating the prevalence of autoimmune diseases. Their study, which included 14,000 participants, found that the incidence of autoimmune diseases in adolescents nearly tripled between 1988 to 2012 ("Autoimmunity in Adolescents Tripled in Recent Decades"). And as can be seen in the anecdotes described in the previous paragraph, it is not uncommon for the victims of the psychogenic inference to be autoimmune diseases patients.

The tendency of doctors to automatically label a patient's medically unexplained symptoms as a psychiatric case is an example of a single story, a concept introduced in Chimamanda Adichie's 2009 TEDx talk, "The Danger of a Single Story". In her talk, Adichie explains that single stories are the base from which stereotypes and norms emerge (13:03). She states that the danger of stereotypes is in their incomplete view in the grand scheme of things (13:06). Adichie eloquently sums up the danger of stereotypes in her statement: "They make one story become the only story" (13:14). She then goes on to identify the power that single stories hold over those whom single stories are about. She claims, "show a people as one thing, as only one thing, over and over again, and that is what they become" (9:14). According to Adichie's statement then, one could argue that the medical norm of psychiatric misdiagnosis in rare disease patients is a type of single story. Undiagnosed rare disease patients are told repeatedly by different physicians the single story that their symptoms are not real but a figment of their imagination.

Dr. Robert Smith, Professor of Medicine and Psychiatry at Michigan State University, provides insight for some of the reasons why the medical field has been perpetuating this single story. Dr. Smith emphasizes the importance of diagnosing patients with severe medically

unexplained symptoms with depression or anxiety. He states that mental disorders are high in the general population and stresses the urgency of appropriate treatment, which can be provided immediately upon a psychiatric diagnosis, but not upon the vague diagnosis of MUS (Smith 1-2). However, Dr. Allen Frances, former chair of psychiatry at the Duke University School of Medicine, stresses the dangers of inaccurate psychiatric diagnoses with this statement: "False certainty is much more dangerous than uncertainty" (Cohen). Patients misdiagnosed with a psychological disorder are often prescribed medications such as antidepressants, which comes with side effects and risks (Pocinki 43:10). Furthermore, inaccurate psychiatric diagnoses in electronic patient medical records can prove problematic due to the difficulty of removing these labels when proven wrong (Pocinki 43:30). And as previously shown in the cases of Professor Atkins and the woman in Japan, false certainty endangers a patient's physical health by delaying crucial medical treatment.

Dr. Frances attributes the tendency for doctors to opt for false certainty to their unease of not being able to provide an answer for the patient. In response to this unease, Dr. Frances states that "doctors need to learn to embrace medical uncertainty" (Cohen). Unfortunately, the medical field ignores the fact that "it is perfectly possible that there is some physical cause present, but that modern science has not yet discovered what it is" (Sykes 291). As a result, undiagnosed rare disease patients are consistently invalidated by their physicians. Rebecca Nunn, a medical student from the University of Cambridge suggests that ironically, these invalidating, stress-filled interactions can result in the development of actual psychological issues such as anxiety and depression in addition to the biological illness that is being dismissed (Nunn 2).

It is also important to acknowledge the significant role that the structure of our medical system plays in maintaining this norm. Medical insurance only provides reimbursement for fifteen-

minute medical consultations (Lee). According to a recent study, physicians typically spend at least a third of the fifteen minutes reviewing a patient's paperwork, which leaves approximately only eight minutes for physical examination and discussion with the patient (Lee). With such a tight schedule, it would not be possible for a doctor to find the time to further investigate puzzling symptoms. It becomes more practical to simply label a patient presenting with MUS as a psychiatric case that can then be referred to the appropriate specialty.

Furthermore, the symptoms of psychiatric disorders and rare diseases sometimes overlap, exacerbating the perpetuation of this norm. Dr. Alan Pocinki, an established rare disease specialist in Maryland, has found that "some apparently psychiatric disorders are instead disorders of adrenaline and the autonomic nervous system" (Pocinki 13:47). The autonomic nervous system controls involuntary functions such as breathing, heart rate, and digestive processes (14:03). In disorders of the autonomic nervous system, patients can have symptoms like extreme fatigue, abnormal heart rate, anxiety, dizziness, and headaches due to a dysfunction of the involuntary regulation of certain hormones in the body (21:45 – 27:52). Dr. Pocinki claims that often the list of symptoms required for diagnosing mental disorders are the same list of symptoms that are present in rare illnesses (36:08). This identical presentation of symptoms combined with the tight time schedule doctors are required to adhere to can provide an explanation for the prevalence of these psychiatric misdiagnoses.

The tendency for doctors to make swift conclusions that result in these misdiagnoses is reminiscent of the historical concept of hysteria. Historically, the word hysteria referred to a mental illness that was thought to predominantly affect the female sex. A large variety of symptoms were associated with hysteria, including but not limited to pain, muscle spasms, paralysis, emotional outbursts, blindness, and hallucinations (Cherry). The concept of hysteria first originated in the 5th

century BCE with the Greek physician Hippocrates, who believed hysteria was a female disease that resulted from a dissatisfied, wandering uterus (Murphy 1806). In more recent history, Sigmund Freud defined hysteria as a predominately female psychological disorder triggered by sexual abuse or trauma that occurred during childhood (McVean). Freud theorized "that the unconscious mind could produce physical symptoms when dealing with memories or emotions too painful for the conscious mind to handle" (Brea 6:54). Paul Briquet, a 19th century French physician and psychologist, established the connection between hysteria and MUS when he claimed that hysteria was "a chronic disorder characterized by the presentation of many medically unexplained symptoms in the body's multiple organ systems" (North 499).

In a 2016 TEDx talk, Jennifer Brea shares her personal experience with MUS that prompted a misdiagnosis of conversion disorder, the modernized medical label that emerged in the 1980s to replace hysteria (Brea 8:55). She was a PhD student at Harvard when she contracted a virus and ran a fever of 104.7 degrees (0:34). In the following days, Brea began to experience constant infections, dizziness, and extreme weakness that at times rendered her physically immobile and unable to speak (1:00-1:59). Her doctor diagnosed her with conversion disorder and informed her that her symptoms stemmed from a distant, emotional trauma that her conscious mind was not aware of (2:18). Eventually, Brea was correctly diagnosed with myalgic encephalomyelitis, a rare and debilitating biological disease colloquially known as chronic fatigue syndrome.

Given that the origin of the norm of dismissing a patient's mysterious symptoms and attributing its cause to a mental disturbance can be linked all the way back to Hippocrates' scientifically inaccurate views regarding hysteria, the current perpetuation of this norm is inexcusable. Not only does this norm imply an unwillingness to do away with the lingering effects of the misogynistic ideologies surrounding hysteria, but also, the anecdotes mentioned throughout

this paper provide evidence that the modern-day perpetuation of this single story inflicts grave physical and mental harm to rare disease patients. In the opinion of Dr. Mark Graber, a professor at Stony Brook University, diagnosing a patient with a mental disorder when no biological cause turns up is not justified until thorough medical investigation has been completed (Cohen).

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