

Commentary on the Practice of Medicine (11): Wheels of Justice Turn Slowly but Grind Exceedingly Fine

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Received October 10, 2023; Revised October 20, 2023; Accepted November 10, 2023

AN EXPECTED SHIFT

The sentence above about the wheels of justice is a metaphor attributed to both ancient Greek and Chinese philosophers. It acknowledges the often slow pace of attaining justice, but advises patience because eventually justice is achieved. In the Portuguese language, the expression "here we do, here we pay" is also used for the sense of justice. It tells us to be careful with our attitudes so we will not regret the consequences. That's the reason why I am very careful with what I write. I don't want to hurt anyone. Essentially, my objective is to defend myself from the allegations made against me, as I believe I was subjected to disrespectful treatment. The authorities and colleagues involved failed to consider my statements, written explanations, and the information I presented regarding my integrity as a medical doctor. To my surprise, a recent statement by a member of the Federal Council of Medicine has heralded a momentous transformation. He said that the resolution no. 2.336/2023 represents a profound paradigm shift within the realm of medicine. The existing regulations, enshrined in laws, he said, have surprisingly been misinterpreted for many years. It was only four years ago, upon revisiting certain texts, that the members of the Council became aware of these misinterpretations, prompting a reevaluation of them all [1]. From my perspective, I had personally foreseen that something would occur sooner or later. The prevailing issue of power wielded capriciously against certain doctors - though not all - without clear justification had left us expecting some change. However, all the accusations against me weathered significant turmoil and endured a lot of damage to both my personal and professional life and I must confess that the promise of change did not immediately offer solace. But it does signify a long-awaited shift, and now, perhaps, apologies could truly bring some comfort.

ANOTHER INARTICULATE ACCUSATION

I guess I could leave it all totally behind now. Nevertheless, I will carry on writing. It is a personal delight in moments when the quietness around me brings me inspiration. Have you read my previous articles? Many accusations were under the same supposedly violated 14th and 18th articles of the

Brazilian Code of Medical Ethics which, in short, say that I did something unnecessary or prohibited in the country. What is truly remarkable is that mere accusations, such as "prescribing genetic tests for personalized dietary prescription", were considered enough, without any additional context. I really don't know what is the issue about these tests. They aren't explicitly prohibited. Therefore, it appears they were deemed unnecessary. This presents an excellent subject to delve into and explore in more depth. Yet, remember that I am just an ordinary clinical doctor. If they are questioning the accuracy of the genetic tests, they should talk to a DNA gene expert. My area of focus is Nutrigenomics, and I heavily depend on the information from databases such as Gene cards, The Human Gene Database, and The Nucleotide Database of the National Library of Medicine. Additionally, I depend on a specialized genome center in my country to obtain comprehensive DNA data from my patients. This center provides a detailed report on over 200 genes, which I then correlate with the patient's medical and family history, lifestyle choices, including diet and physical activity, symptoms, and laboratory test results. This intricate process enables me to make more accurate interventions aimed at enhancing longevity and promoting better health along the process of aging than previous generations - what is something desirable for most of us, if not all.

GENETIC TESTING

It is widely known that genetic testing looks for changes, sometimes called mutations or variants, in our DNA. The Center for Disease Control and Prevention (CDC) states in

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Citation: Stoop IMB. (2024) Commentary on the Practice of Medicine (11): Wheels of Justice Turn Slowly but Grind Exceedingly Fine. BioMed Res J, 8(1): 651-654.

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their site that "genetic testing is useful in many areas of medicine and can change the medical care you or your family member receives" [2]. They hold a significant value in the field of science, continually advancing and gaining credits in the last decades, mainly after 2003 when the Human Genome Project was completed [3]. Here are some reasons which highlight the importance of genetic testing:

- Genetic testing has been crucial for diagnosing genetic disorders and assessing an individual's risk for developing certain diseases, such as hereditary cancers, rare genetic diseases and predispositions to different conditions [4,5] like Alzheimer's and Celiac Disease.
- Genetic testing has played a pivotal role in the advancement of personalized medicine.

By analyzing the patients' DNA, healthcare providers can tailor dietary plans and treatments to an individual's unique genetic profile, improving efficacy, minimizing adverse effects and favoring well-being and better health in the long run [4,6].

- Genetic testing has been used to assess the risk of passing on genetic disorders to offspring, helping couples to make informed decisions about family planning and reproductive options [4].
- Genetic testing has been instrumental in scientific research, helping scientists better understand the genetic basis of various diseases and traits. This information is vital for advancing our knowledge of genetics and developing new therapies and treatments [4].
- Genetic testing contributes to population epidemiological studies by identifying genetic variations associated with specific health outcomes. This information can be used to create public health interventions and strategies [7,8].
- Genetic testing techniques, such as DNA profiling, have been invaluable in forensic science for identifying individuals, solving crimes, and establishing paternity [9,10].

PRECISION NUTRITION

In my area of expertise, a 2023 study points out that "precision nutrition is an emerging area of nutrition research that focuses on understanding metabolic variability within and between individuals and helps develop customized dietary plans and interventions to maintain optimal individual health. It encompasses nutritional genomic (gene-nutrient interactions), epigenetic, microbiome, and environmental factors" [11]. As far as dieting is concerned, it's already known that not only energy in terms of calories,

but also the specific composition of the diet can affect the way the food is absorbed and how its energy is stored, used or dissipated [12]. Also, one thing is to offer the established amounts of nutrients considered essential to attend our basic needs. Another approach is to offer extra protection, including active compounds that seem to act effectively in the hallmarks of aging [13-16]. It's important to acknowledge that this field is relatively new, and there's a considerable journey ahead of us. In this regard, I consistently thank all my patients for participating in the testing process, emphasizing that they are actively contributing to the advancement of science as a whole and the refinement of genetic testing specifically. I make it clear that meaningful progress can only be achieved through our collective commitment to thoroughly investigate and apply the insights derived from these tests.

AN INTERESTING AND COMPLEX GENE MUTATION

I have always been intrigued by high Blood Nitrogen Urea (BUN) results with normal levels of creatinine in well hydrated patients, with no evidence of liver diseases, kidney problems or excessive protein intake. Through genetic testing, I have learned about what is known as cystathionine beta-synthase (CBS) gene mutation, or CBS mutation. In fact, according to the Human Gene Mutation Database, more than 200 pathogenic mutations in the CBS gene have been identified worldwide [17], but one, in particular, causes the CBS enzyme to work too fast (elevated activity). CBS is the first step of the transsulfuration pathway, converting homocysteine to cystathionine [18]. The transsulfuration pathway, also known as the liver pathway, is a metabolic pathway where transfer of sulfur from homocysteine to cysteine occurs [19]. It leads to either taurine or glutathione production [19]. In this case, homocysteine and cystathionine convert too quickly to taurine, leaving not enough glutathione for the body. As the most vital antioxidant and normally the most abundant, glutathione is required for many processes, including immune function, detoxification, and the function of other antioxidants [20]. Without enough glutathione, very little is filtered and processed in your body, leaving it functioning abnormally. It has also been observed that tetrahydrobiopterin (BH4) [21], an enzyme that is used to make serotonin [22], dopamine [22], nor-epinephrine [22], nitric oxide [23,24] and melanin [25], in addition to helping the detoxification of ammonia from the body, can also become depleted with the CBS mutation. BH4 is extremely important as it has multiple roles in mood and neuropsychological disorders [22]. Other gene mutations, such as methylenetetrahydrofolate reductase (MTHFR) A1298C, chronic bacterial infections, and aluminum, can also lead to low BH4 levels. Also, the CBS upregulation substantially upsets the urea cycle: this enzyme depletes the BH4 enzyme while overloading it. And, finally, the CBS mutation may lead to an excess amount of taurine and

sulphur in the system. It may therefore be highly recommended that, if you have this mutation, you limit your sulfur intake as it can create excessive stress on your system. Complex sulfur, such as in horsetail grass, spirulina, dandelion leaf, or parsley, is usually fine. For this reason, cravings for sulfur, an aversion to it, or a sensitivity to sulfur-rich foods may warrant investigating the possibility of a CBS upregulation. In this case, sulphur-rich foods (broccoli, cauliflower, kale, garlic, onions, cabbage, watermelon and sweet potatoes) and supplements (biotin, lipoic acid, N-acetyl-L-cysteine, L- glutamine, L-taurine, S-adenosyl-L-methionine, methyl-sulfonyl methane or MSM, chondroitin sulphate, magnesium sulphate, milk thistle, di-mercapto-L-propane-sulphonic acid or DMPS) must be ingested with some restriction [26]. Driven by curiosity, I randomly selected thirty of my patients who had undergone Nutrigenomics Panel testing. Among them, I found out that fourteen exhibited a homozygous mutation in the CBS gene, fifteen had a heterozygous mutation, while only one patient had no mutation in the same gene. This finding has ignited my enthusiasm to delve deeper into the study of CBS gene mutations, as learning what to do holds the potential to significantly enhance the health of numerous patients. By optimizing some detoxification pathways and bolstering overall well-being, I anticipate that this research will serve as a prime illustration of how genetic testing can be a valuable asset in my role as a clinical nutrition practitioner. It empowers me to provide more personalized, precise dietary recommendations and supplement prescriptions.

THE SACRED PATH OF BEAUTY

A documentary on television is currently highlighting the concept of "Blue Zones," regions around the world where a significant number of people live to be over a hundred years old [27]. The Blue Zones include Okinawa in Japan, Ikaria in Greece, Sardinia in Italy, Nicoya in Costa Rica, and Loma Linda in the United States. At some point, more to the end, it mentions life in Singapore. Spanning just about 275 square miles, this city, nation and state is very small, but, yet, it's home to a population of five million people. Despite its compact size, it appears to boast a notably high quality of life and the highest life expectancy in the world today. What surprised me the most, however, was what Chan Hang Chee, Ambassador-at-large and Minister of Foreign Affairs, said: "We try to help people to help themselves". That's exactly what I do with my work: I try to guide my patients to find their own path of cure. That reminded me of Socrates, the renowned Greek philosopher who never penned a single word, but is still revered as one of the greatest thinkers in history. He used to say that he didn't want to teach anything to anyone. He wanted to help them to find their own solutions to their problems. In other words, according to him, the discernment has to come from within. My point is: when we engage in actions aligned with what we genuinely perceive as the "right thing", our brain validates these emotions, resulting in a sense of well-being. As simple as

that. There will be no reward feeling inside ourselves otherwise. I firmly believe that we should adopt this as a guiding principle in our everyday lives, akin to what the American indigenous people advocate as "the sacred path of beauty": the journey in which we harness our talents to heal ourselves and nurture the Earth. With love and glory.

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