Precision dinner: Genomes and health records

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Did personalized genome sequencing help me? ‘Fast caffeine and alcohol metabolizer’ was not impressive. I found a LoF (Loss of Function) mutation in Coagulation Factor X (F10).It was fortunate that it was heterozygous, as homozygosity might have prevented my birth. My factor assay revealed that my F10 functional activity was at 67%, which is below normal, along with delayed BT, PT and aPTT. Although I am a medically proven F10 deficiency patient, I have no bleeding tendency. Perhaps I am more evolved in this era of hypercoagulability due to our civilization progressing faster than evolution. The problem is that tens of millions of people are prescribed anticoagulants to prevent cerebrovascular accident. Equipped with the knowledge of my genome and its mutations, I know that I should not take anticoagulants. However, the main indication of F10 inhibitors has nothing to do with coagulopathy nor clotting. The prescription of anticoagulants for atrial fibrillation is common, and doctors without the knowledge that I have a LoF variant that would make my response to the drug dangerous would not hesitate to prescribe them to me, or to other patients like me. There is no rationale for prescribing an inhibitor for a target that has already been inhibited by LoF variants. More than 300,000 Koreans (1/167) are at risk due to this specific mutation. Xantine Dehydrogenase (XDH) inhibitors are treatments of choice for gout but may be useless for me due to another LoF variant present on my XDH. Distal tubule reuptake inhibitors may be alternatives for the one in ten patients with this variant, such as myself. LoF’s can sometimes protect us in mysterious ways, as well. Despite my very mild symptoms, I am self-medicating myself with an endogenous substance with no metabolite nor side effects. I will eventually lose my sight due to a LCAT deficiency, which is causing cholesterol deposits in my lens, and a GALK1 deficiency which is causing cataracts. I have to enjoy my current sight and see all the beautiful things I can on Earth. This kind of information is not outlined in current medical textbooks. Conventional medicine focuses on the average, and considers us all as average folks. Mere “idiosyncrasies” cannot prevent doctors from prescribing potentially harmful medications without guilty in virtue of medial utilitarianism. Will this approach still hold in thisera of digital genomes? We all are minorities in some conditions and against some drugs. One does not fit all, we are all different in some aspects. Medicine has not just been fully personalized yet. Shall we take a walk for precision dinner?