COLLEGE OF NURSING
UNIVERSITY OF THE PHILIPPINES MANILA

3 June 2019

[redacted]

Dear [redacted]:

This is a recap of what transpired during our genetic counseling session last [redacted]

You were referred to me by [redacted], who has previously seen your sister [redacted] because of early-onset breast cancer and multiple cancers in your family history. Genetic testing was done with [redacted] and the results detected a pathogenic variant in TP53 gene that is consistent with Li Fraumeni Syndrome.

I understand that your sister has informed you about the results of the genetic test and she informed you about the possibility that you too can undergo genetic test to determine whether you have inherited the pathogenic variant. The genetic counseling session we had last [redacted] was important in order for you to understand the risks and benefits of genetic testing so that knowing these, you can make an informed decision whether to undergo the test or not.

Li Fraumeni Syndrome (LFS) is one type of a cancer syndrome that is inherited in the family. Majority of the cases of LFS are caused by mutations in the TP53 gene, an important gene that regulates division of cells in the body. When a car is used as an analogy, the TP53 gene acts like the 'break' that stops cell division. Without this 'break', cells will continue to divide unregulated which is a general feature of cancer. The TP53 gene mutation can be inherited in the family usually in a pattern we call *autosomal dominant*. In this pattern, only one copy of the mutated gene (we have two copies of each gene—we inherited one copy from the mother, and another copy from the father) is enough in order for LFS to manifest and that individuals with the mutation has a 50% probability of passing on the mutation to their children. In other words, children of parents with TP53 gene mutation has 50% chance of inheriting the mutation. In the same token, brothers and sisters of someone who has TP53 gene mutations also have 50% chance of having the same mutation. The only way to determine if a person has inherited the mutation is through genetic testing.

Li Fraumeni usually is associated with various cancers most notably sarcomas (which are cancer of connective tissues like bone and cartilage), breast cancer, adrenal cancer, and pancreatic cancer. Individuals who have inherited the TP53 gene are at higher risk of developing the cancers mentioned compared to someone who do not have a mutation in his/her TP53 gene.

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During the session, we also discussed how the genetic testing is done and what could be expected from the test particularly the results. To recap, in case you consent to undergo genetic test, the testing will focus on finding the specific mutation that was found in your sister, [redacted]. This is called a targeted mutation analysis because we are only looking for a specific mutation that is inherited in your family.

The possible results of the genetic testing include a positive and a negative result. A positive result mean that you have inherited the TP53 mutation, the same mutation as with your sister. This means that you have a higher risk of developing LFS-associated cancers in your lifetime and this require rigid surveillance mechanisms in order to detect cancer in its early stages. A negative result, on the other hand, will tell us that you did not inherit the TP53 gene mutation and that your risk of developing LFS-associated cancers is the same as that of an individual from the general population.

Knowing your mutation status through genetic testing may provide very useful information on how to manage your health in the future. This may make you in control of your health. A positive result may mean that you will need to be managed by an oncologist in order to detect cancers early on. But please do understand that this may also increase anxiety and stress for some individuals. A negative result, on the other hand, may ease anxiety of people but for some this may put guilt upon them knowing that they do not have the familial mutation. Please take this time to think about pursuing the genetic test and how you may potentially react with the possible results.

If you have any questions, you may get in touch with me at [redacted].

If you decide to pursue genetic testing, you may present this letter as proof that you underwent genetic counseling and that you have understood the nuances of the test.

Thank you so much.

Sincerely,

[redacted]