Dear ------

I write to inform you that I have recently undergone genetic testing and have been diagnosed with Birt-Hogg-Dubé syndrome (BHD).

BHD is a rare hereditary disease which means it is likely to run within our family, and you and other family members may have this condition.

Research indicates there are currently around 600 families across the world who have BHD, however it is thought that this figure is likely to be a lot higher. The reason for this is that the condition is relatively unknown in the medical profession. So if you visit your GP with any of the below listed symptoms it is highly unlikely that your GP would automatically refer you for genetic testing, and BHD would not show up on any standard checks such as a blood tests.

If BHD is managed well, people with this condition normally lead a normal life. Some of the more common symptoms of BHD include:

* Pale skin bumps normally found on the face, ears, neck, and upper body. These skin bumps are non-cancerous skin tumours and although they are not dangerous, they can cause some people to feel anxious or self-conscious.
* 8 in 10 people with BHD develop lung cysts, and 1 in 4 people have at least one episode of collapsed lungs (pneumothorax). These are not normally life threatening if carefully monitored and some small day-to-day adjustments are made.
* BHD syndrome can cause kidney cysts and kidney cancer. In most cases, tumours within the kidneys grow very slowly, rarely spread, and are not life-threatening so long as they are appropriately treated.

BHD is caused by mutations in the Folliculin gene, and the only definite way to know if you have it is to have a genetic test. The mutation I have is called insert mutation, which is the mutation you might have too. I recommend that you and other family members arrange to have a genetic test to assess if you have this condition. I also recommend taking a copy of this letter to your GP so they know which genetic test to order.

In addition, the BHD Foundation supports individuals and families who have BHD. They also collaborate with the Myrovyltis Trust to provide funding for research into BHD, and to educate clinicians on the symptoms, diagnosis, and treatment for patients. For further information please visit thebhdfoundation.org.

If you would like to talk to me about the above, please do contact me on ------ or email me at ------.

Best wishes

------