

# Rare Disease Day 2017



As rare disease day comes around again I thought I'd take a minute to talk about rare diseases, what they are, what common problems occur and what characteristics they carry.

I am also going to share a rare disease that many people are unaware about but one that has affected someone close to my heart.

## **So, what is a rare disease?**

A disease or disorder is classed as rare when it affects a small percentage of the world's population. In Europe, a disease or disorder is classed as rare when it affects as little as 1 in 2000 people.

***"One rare disease may affect only a handful of patients in the EU (European Union), and another may touch as many as 245,000. In the EU, as many as 30 million people may be affected by one of over 6000 existing rare diseases."*** -

<http://www.rarediseaseday.org/article/what-is-a-rare-disease>.

## **What are the characteristics of a rare disease?**

There are over 6000 rare diseases that are characterised by a variety of symptoms and disorders that vary from disease to disease. These characteristics can also vary from patient to patient that are suffering from the same disease.

However, some of the most common symptoms such as a headache, dizziness and tiredness also hide underlying rare diseases that can lead to a misdiagnosis and a delay in treatment. This affects the patient's quality of life due to the threatening traits of the disease.

***"80% of rare diseases have identified genetic origins whilst others are the result of infections (bacterial or viral), allergies and environmental causes, or are degenerative and proliferative."***

- <http://www.rarediseaseday.org/article/what-is-a-rare-disease>.

## **Some common problems that occur with rare diseases?**

There are many common problems that can occur when it comes to rare diseases. The most common problem is the lack of scientific knowledge. Many doctors and scientists are always trying to proactively work towards new defining and treating rare diseases. However this is a long process involving many years of research and trials and in many cases this is too long to prevent many people losing their life.

## Mycobacterium Xenopi.

Mycobacterium Xenopi was first discovered in 1959; it originates from a South African toad and is recognised in patients that suffer from chronic lung disease. Xenopi attacks the weakest part of the body, in this case it attacks the lungs as they are weak from the lung disease. It attacks by eating away at the organ.

Mycobacterium Xenopi is a Fungi that lives in the body for a while showing no symptoms, occasionally it can fluctuate and after lying dormant in the body for a long period of time can cause serious difficulties. Many doctors are unaware of Xenopi leading to a delay in diagnosis.

Back in 2009 my Grandfather passed away from xenopi. Doctors where baffled on how he developed the disease as it's not common in the UK especially as he had never travelled out the country.

Doctors later discovered that he may have picked it up from having the lining of his lung removed when he was younger, or he picked it up from someone he may have met whilst working in the police force.

The symptoms of Xenopi are breathing difficulties, loss of appetite, weight loss, extreme tiredness and weakening to the muscles.

There is no treatment for Xenopi, however many cases have led to having an organ transplant. Even then Doctors are sceptical about how successful transplants can be as in many of these cases the Fungi can also attack the new organs.

***“Mycobacterium xenopi is considered infectious but is not transmitted from person to person. Generally, a disease like this is caused by an infectious agent and not spread between people.***

***Mycobacterium xenopi, although infectious, is not a genetic disease. It is not caused by a defective or abnormal gene.” -***

[http://www.rightdiagnosis.com/m/mycobacterium\\_xenopi/contagious.htm](http://www.rightdiagnosis.com/m/mycobacterium_xenopi/contagious.htm)

Till this day many doctors are still unaware of xenopi and the worldwide research continues.