RARE #RARENESSAWARENESS

FAQ's

WHAT IS A RARE DISEASE?

There is no universal definition of a rare disease (RD) and they are defined differently in different countries and regions:

* In the EU, RD were defined as conditions with a prevalence affecting less than 1 in 2000 (EU Regulation on Orphan Medicinal products (1999).
* In the USA they are defined as a disease affecting less than 200 000 people in the country (American Orphan Drug Act of 1983). Other countries range from 5-76 per 100 000 of the population.
* South Africa has adopted the European definition of RD – for less than 1 in 2000 people affected.

HOW MANY RARE DISEASES ARE THERE?

To date 7,000 rare diseases have been described. It is estimated that there are 350 million people living globally with a rare disease.

In South Africa, it is estimated that there are 1 in 15 affected, equating to 4.1 million people living with a rare disease.

Current South African laws do not require rare diseases to be reported. While many infectious diseases (e.g. HIV/AIDS, TB, rabies, cholera, COVID19 etc, and all cancers are reported, the only congenital disorder/birth defect that is currently notified in South Africa is congenital syphilis.

Because rare diseases are not counted in South Africa, the total number of people affected by rare diseases is unknown. This makes it impossible to plan health care services for the care and management of rare diseases.

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HOW AND WHEN ARE RARE DISEASES DIAGNOSED?

It takes an average of 6-8 years to get a diagnosis for rare disease. During this "diagnostic odyssey" patients may be misdiagnosed several times before they receive a correct diagnosis and are referred for appropriate care.

Many rare diseases share symptoms with more common ailments – which Drs are trained to look for first "*If you hear hoofbeats look for horses not zebras"*.

While some RD are obvious, many are invisible, making them more difficult to diagnose, such as inborn errors of metabolism and many congenital heart defects. An early and accurate diagnosis enables relevant, timely treatment. This may be lifesaving and/or slow progression of the disease and improve quality of life significantly.

Comprehensive Newborn Screening (NBS), screening of newborns for specific conditions in many countries, is not available in state health services in South Africa. In other countries, over 50 conditions are screened for, saving lives and mitigating disability. In some cases, treatment of these conditions is not expensive, such as congenital hypothyroidism for which treatment only costs R30 per month. Other conditions only require a specific diet to be followed.

Because there are so many rare diseases (>7000) many Drs (e.g. GPs) do not have clinical experience or expertise in rare diseases. There is a lack of knowledge for many rare diseases and while research is increasing, it takes time for this to be applied in health care.

While diagnostics of rare diseases are improving due to new emerging technologies, it is not enough to save many patients who may not survive to receive a correct diagnosis.

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WHAT ARE SOME EXAMPLES OF RARE DISEASES?

Rare diseases are present across the medical spectrum. Some are widely recognized by name, such as cystic fibrosis, while others are less known, such as cat eye syndrome.

Most cancers (all but a few types) are rare. There are rare neurological and neuromuscular diseases, metabolic diseases, chromosomal disorders, skin diseases, bone and skeletal disorders, and rare diseases affecting the heart, blood, lungs, kidneys, and other body organs and systems.

Many rare diseases are named for the physicians who first identified them. A few are named for patients or even the hospitals where they were first identified.

While rare diseases may be found in every population in the world, some are considered rare in some countries and more common in other countries. This is because the prevalence of rare diseases may differ between different population and ethnic groups.

WHAT CAUSES RARE DISEASES?

The majority of rare diseases (70-80%) are genetic, caused by changes in genes or chromosomes.

In some cases, genetic changes that cause disease are passed from one generation to the next (inherited). When these inherited changes occur for the first time in families they are called “de novo” or new changes. In other cases, random genetic changes may occur before conception causing a disorder in the individual only, such as most cases of Trisomy 18 (Edward’s syndrome).

Many rare diseases, including infections, some rare cancers, some autoimmune diseases, are not inherited and may be caused by interaction with the environment. While researchers are learning more each year, the exact cause of many rare diseases is still unknown.

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WHAT IS BEING DONE TO DEVELOP TREATMENTS FOR RARE DISEASES?

Researchers have made progress in learning how to diagnose, treat, and even prevent a variety of rare diseases. However, there is still much to do because 90% of rare diseases have no FDA approved treatments.

Efforts to improve and bring to market treatments for rare diseases are coordinated by the Food and Drug Administration (FDA). The Office of Orphan Products Development (OOPD) provides incentives for drug companies to develop treatments for rare diseases.

Between 1973 and 1983, fewer than 10 treatments for rare diseases were approved. Since 1983, the OOPD program has helped develop and bring to market more than 800 drugs and biologic products for rare diseases.

For rare diseases where there is no approved treatment, other medications may be used 'off label'. This means that while they were not developed

WHAT ARE SOME OF THE PROBLEMS PEOPLE WITH RARE DISEASES EXPERIENCE?

* Difficulty in obtaining an accurate diagnosis (this can take years, which can be critical for stopping or halting the progression of a disease)
* Limited treatment options
* Little or no research being done on the individual raredisease
* Difficulty finding physicians or treatment centres with experience/expertise for a particular disease
* Treatments may be more expensive than those for common diseases
* Reimbursement issues related to private insurance and out of pocket payments
* Difficulty accessing disability or social, grants because those making the decisions are not familiar with the disease
* Feelings of isolation and of having been abandoned or "orphaned" by our health care system

FAQ's

* Lack of awareness and support in the community

WHAT IS RARE DISEASE DAY?

Rare Disease Day is an awareness event that takes place every year on the last day of February, February 28 or February 29 in Leap Years—the rarest of calendar dates to underscore the nature of these diseases—to focus public attention on rare diseases as a public health concern.

WHEN WAS RARE DISEASE DAY STARTED?

Rare Disease Day was first observed in Europe in 2008. It was established by EURORDIS, the European Rare Disease Organization. In 2009, EURORDIS asked NORD to be its partner in this initiative and to sponsor Rare Disease Day in the United States.

RDSA first participated in 2010.

WHAT HAPPENS ON RARE DISEASE DAY?

Patients, their families, caregivers, researchers, advocates and others get involved through storytelling, media interviews, posting stories, videos and blogs online, hosting or attending events, and educational initiatives in classrooms and on college campuses. A wide range of activities are planned and executed to celebrate the day.

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