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bones [2] genetics [3] lungs [4] disability [5]

Rare diseases, or rare conditions as they are sometimes known, affect fewer than 1 in 2000 people in the UK, meaning that some people face their rare journey alone. Globally, as many as 446 million people live with one of these conditions. Some, such as cystic fibrosis, are well-known; others don't even have a name. Thanks to work by charities such as Beacon, rare conditions are starting to receive the visibility, awareness and research they deserve.

There are more than 6,000 rare diseases. Blayne Baker, Marketing and Engagement Manager at Beacon, highlights seven contrasting pairs to show how different they are in terms of their cause, symptoms and impact.

Friedreich's ataxia and Guillain-Barré syndrome

A **genetic** condition that's inherited from a parent, Friedreich's ataxia causes poor balance and coordination, difficulty speaking and swallowing, reduced sensation in one's limbs and trouble handwriting. Those living with Friedreich's ataxia often appear drunk [6] due to neurological symptoms.

72% of rare diseases are genetic in nature, being caused by an error or mutation in a person's DNA that changes the way the body functions. Others are caused by infection, allergy or environmental causes such as diet, smoking or exposure to chemicals. A condition that's triggered by an **infection** as common as the flu or food poisoning is Guillain-Barré syndrome [7] (GBS). This rare condition causes a rapid onset of numbness, weakness and often paralysis in the face, arms, legs and breathing muscles.

Alkaptonuria and Fibrodysplasia ossificans progressiva

Rare diseases can touch a person's life at any moment. Alkaptonuria [8] (more commonly known as AKU or Black Bone Disease) affects people **later in life**. Those living with AKU produce black urine and experience significant damage to their bones and cartilage. In adulthood, AKU patients may experience early-onset osteoarthritis, multiple joint replacements and heart valve problems.

The rare condition Fibrodysplasia ossificans [9]progressiva [9] (FOP) touches a person's life at the **moment of conception**. FOP is caused by a spontaneous mutation early in the development of an embryo. It is one of the rarest and most disabling genetic conditions known to medicine. It is the only known disease where one body system turns into another. Soft tissue turns into hard bone, creating a second skeleton. It significantly shortens a person's life expectancy.

Achondroplasia and Myasthenia Gravis

Rare diseases can be **immediately visible** to others. Achondroplasia [10] (dwarfism) is a visible skeletal disorder that affects bone growth and can be either inherited from parents who have the condition or caused by a mutation in the embryo as it forms. Those living with achondroplasia have short stature, an enlarged head with a prominent forehead and limited range of movement in their elbows.

But many rare conditions are **less visible** to the eye. Vascular Ehlers-Danlos syndrome [11] causes a weakness in the walls of the arteries. Subtle signs of the disease can include varicose veins at a young age and easy bruising from birth. Myasthenia Gravis [12] affects the muscles that control the eyes, eyelids, facial expressions, chewing, swallowing and speaking.

Duchenne Muscular Dystrophy and Turner Syndrome

Rare diseases don't discriminate. They can affect anyone, but some specifically affect **one gender**. Duchenne Muscular Dystrophy [13] (DMD) is a genetic condition that's caused by a mutation in the dystrophin gene. It affects 1 in 3,500 boys worldwide, and is very rare in females.

A rare condition that **only affects females** is Turner Syndrome [14]. Affecting 1 in 2,000 females, it's caused by the complete or partial deletion of the X chromosome (one of the two sex-determining chromosomes). The syndrome results in short stature and infertility.

Cystic fibrosis and retinitis pigmentosa

The trouble with rare conditions is that they usually don't just affect one body system. Cystic fibrosis [15] affects **many organs**, including the lungs, intestines and pancreas. When rare diseases affect multiple organs and systems, they can be harder to diagnose and treat: key connections between symptoms that could lead to a prompt and correct diagnosis often go unmade.

There are rare conditions affecting **one particular body system**. Retinitis pigmentosa [16] (RP) affects the retina, which is located at the back of the eye. This condition slowly stops the retina from functioning, leading to loss of vision, changes to peripheral vision or altered central vision depending on age and severity.

SWAN and Dubin Johnson Syndrome

Some rare medical conditions remain **unexplained and unnamed**. Syndromes without a name (SWAN) are genetic conditions that are so rare that they often go undiagnosed. Approximately 6,000 children in the UK are born each year with one. SWAN UK [17] is the only dedicated support network for SWAN families in the UK who are fighting for a genetic diagnosis for their child.

Occasionally, rare conditions **are named** after the physician who first identified the disease. An example is Dubin [18] Johnson syndrome [18], a rare condition affecting the liver, described by Dr Frank Johnson and Dr Isadore Dubin in 1954.

Spinal Muscular Atrophy and 95% of rare diseases

The sad reality is that 95% of rare conditions do not have a treatment.

Thankfully, Spinal Muscular Atrophy (SMA) is now treatable. Spinal Muscular Atrophy is a fatal genetic disease,

causing paralysis, muscle weakness and a loss of movement. The one-off gene therapy, Zolgensma [19], said to be the 'world's most expensive drug', is now available on the NHS. It's estimated that 80 babies and young children could benefit from this life-changing gene therapy each year.

Challenges of living with a rare condition

Misdiagnosis, delayed treatment, poor care coordination and a lack of support are common when it comes to rare conditions. Having a rare disease can be an isolating experience for the patient and their family. Rare Disease UK [20] (RDUK) is the national alliance for people with rare diseases and all who support them. According to their survey [21]:

- The average rare disease patient waits 4 years before receiving a final diagnosis [22] after receiving 3 misdiagnoses and consulting with 5 doctors.
- 2 in 3 rare patients and carers struggle to hold paid employment.
- 3 in 5 rare patients feel that their rare disease affects their education.

Patients have felt worried, anxious, emotionally exhausted or at breaking point.

A 'beacon' of hope

Beacon for Rare Diseases is a UK-based charity that is building a united rare disease community with patient groups at its heart. The charity has just celebrated its 10th year!

Patient groups can be turned to for credible information, understanding and support. Sadly, only half of the known rare conditions have one. That's why Beacon offers free training [23] to help these often small and voluntary organisations to form and grow, so that they can become active partners in research and advocacy.

If more patient groups have the opportunity to connect and collaborate with each other, fewer patients will face their rare journey alone.

MEDICAL JOURNALISTS' ASSOCIATION

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