

# RARE DISEASES

World Health Organisation: Rare diseases are a global health priority for equity and inclusion

1 in 2000  
AFFECTED



## What is a rare disease?

- ❖ A specific health condition affecting 1 in 2000 individuals or fewer in the general population
- ❖ There are currently over 7000 known rare diseases impacting more than 300 million people globally, with 70% of these conditions starting in childhood
- ❖ They are often complex and multisystemic, affecting multiple organs
- ❖ They often lead to comorbidities
- ❖ Many of these conditions are chronic, progressive, and can consequently result in serious disabilities and premature death
- ❖ Reaching the correct diagnosis can take over five years, so many persons living with a rare disease never receive a timely or adequate diagnosis

Examples of Rare Diseases which might be seen in the UK working population:



Diseases that someone is born with:

### Neurofibromatosis type 1 (NF1)

- ❖ Prevalence: ~1 in 3,000
- ❖ Genetic from birth; often mild; highly variable symptoms
- ❖ [Click here for more](#)

### Marfan syndrome

- ❖ Prevalence: ~1 in 5,000
- ❖ Genetic from birth; connective tissue disorder; often diagnosed in adolescence/adulthood
- ❖ [Click here for more](#)

### Huntington's disease

- ❖ Prevalence: ~1 in 10,000
- ❖ Genetic from birth; adult onset
- ❖ [Click here for more](#)

### Hypertrophic cardiomyopathy (genetic forms)

- ❖ Clinically significant congenital disease: ~1 in 2,000
- ❖ Often detected via family screening
- ❖ [Click here for more](#)

### Neurofibromatosis type 2 (NF2)

- ❖ Prevalence: ~1 in 25,000
- ❖ Genetic from birth; symptoms often later in life
- ❖ [Click here for more](#)

### Ehlers-Danlos syndromes (classical, vascular, rare types)

- ❖ Prevalence: Each subtype ~1 in 10,000
- ❖ Hypermobile EDS not included due to uncertain prevalence
- ❖ [Click here for more](#)

### Phenylketonuria (PKU)

- ❖ Prevalence: ~1 in 10,000
- ❖ Universal newborn screening in the UK
- ❖ [Click here for more](#)

### Long QT syndrome (congenital)

- ❖ Prevalence: ~1 in 5,000 - 7,000
- ❖ [Click here for more](#)

## Diseases which are acquired during life:

### Mesothelioma

- ❖ Exposure: Asbestos
- ❖ UK prevalence: ~1 in 10,000 (lifetime risk higher in older men, still rare overall)
- ❖ Why rare: Requires heavy historical exposure + long latency
- ❖ [Click here for more](#)

### Farmer's lung (chronic hypersensitivity pneumonitis)

- ❖ Exposure: Mouldy hay/silage
- ❖ Prevalence: <1 in 10,000
- ❖ Why rare: Requires repeated sensitising exposure
- ❖ [Click here for more](#)

### Legionnaires' disease

- ❖ Exposure: Water systems
- ❖ Prevalence: ~1-2 per 100,000 annually
- ❖ Lifetime prevalence: <1 in 2,000
- ❖ [Click here for more](#)

### Isocyanate-induced occupational asthma

- ❖ Exposure: Spray paints, foams
- ❖ Prevalence: <1 in 2,000
- ❖ Note: Severe but rare; high individual risk in exposed workers
- ❖ [Click here for more](#)

### Asbestosis

- ❖ Exposure: Asbestos
- ❖ Prevalence: Well below 1 in 2,000
- ❖ Note: Far rarer than pleural plaques
- ❖ [Click here for more](#)

### Chronic lead poisoning (clinically significant)

- ❖ Exposure: Old housing, hobbies, renovation
- ❖ Prevalence: <1 in 2,000
- ❖ Note: Low-level exposure is more common; disease is not
- ❖ [Click here for more](#)

### Leptospirosis (Weil's disease)

- ❖ Exposure: Rat urine, freshwater
- ❖ Prevalence: ~0.1-0.2 per 100,000 annually
- ❖ [Click here for more](#)

**FEB 28**  
**RARE**  
**DISEASE**  
**DAY**