Challenges in rare diseases

Disease education

Misdiagnosis and missed diagnosis

It can take years to get the right diagnosis:

- Frequent **misdiagnosis** with more common/likely diseases
- Low **awareness** of possible rare diseases
- Lack of **knowledge** of any symptom clusters
- Devastating **impact** on the patient and their family

Patient identification

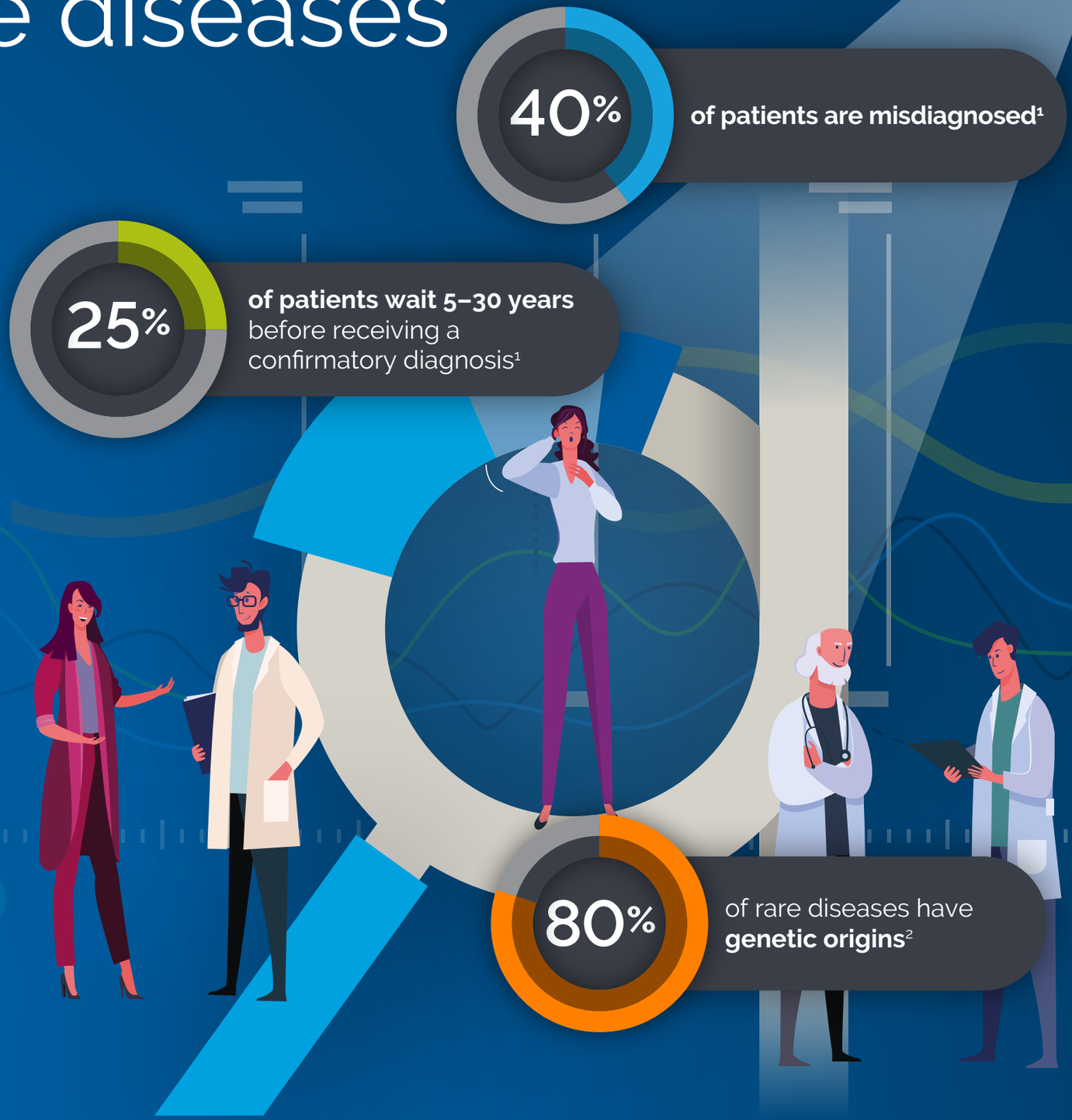
Lack of common approach to patient identification:

- Lack of clarity around **what to look for**
- **No standardisation** of symptom clusters as different patients with the same disease may present with different symptoms
- **Inadequate estimates** of the burden of rare diseases

Patient care pathway

Uncertainty for healthcare professionals and patients after diagnosis:

- Limited **support networks**
- Unclear **referral pathways**
- Lack of guidance on how best to **support patients** and other family members who might be affected **after diagnosis**



1. EURORDIS. Survey of the delay in diagnosis for 8 rare diseases in Europe ('EurordisCare 2'). 2007. https://www.eurordis.org/sites/default/files/publications/Fact_Sheet_Eurordiscare2.pdf. Accessed 3 February 2020;
2. EURORDIS. What is a rare disease? 2007. https://www.eurordis.org/sites/default/files/publications/Fact_Sheet_RD.pdf. Accessed 3 February 2020