Challenges in rare diseases
Treatment

Misplaced treatment expectations
Challenges to find the right treatment (if any):

- Limited **number** of effective treatments
- Lack of meaningful, **real-world evidence** or clear treatment guidelines
- Treatment may **only target symptoms**, not cure

Access
Treatments may be available but not accessible:

- Reimbursement challenges for treatments that are considered too **costly** compared with perceived value of treatment
- Challenging **treatment regimens** for patients
- Limited **access to treatment centres** or specialists

Demonstration of real-life benefits
Reservations around initiating or continuing therapy:

- Lack of **real-world evidence** on patient outcomes and improved QoL
- Concern for potential therapeutic **side effects**, leading to lower patient satisfaction compared with no treatment
- No **perceived benefit** after treatment commencement
- Perception of patient being 'cured' or **no longer needing** expensive ongoing treatment

95% **of rare conditions** have no licensed treatment in the UK[1]

QoL, quality of life
1. Slade A, Isa F, Kyte D, *et al*. Patient reported outcome measures in rare diseases: a narrative review. *Orphanet J Rare Dis* 2018;13(1):61