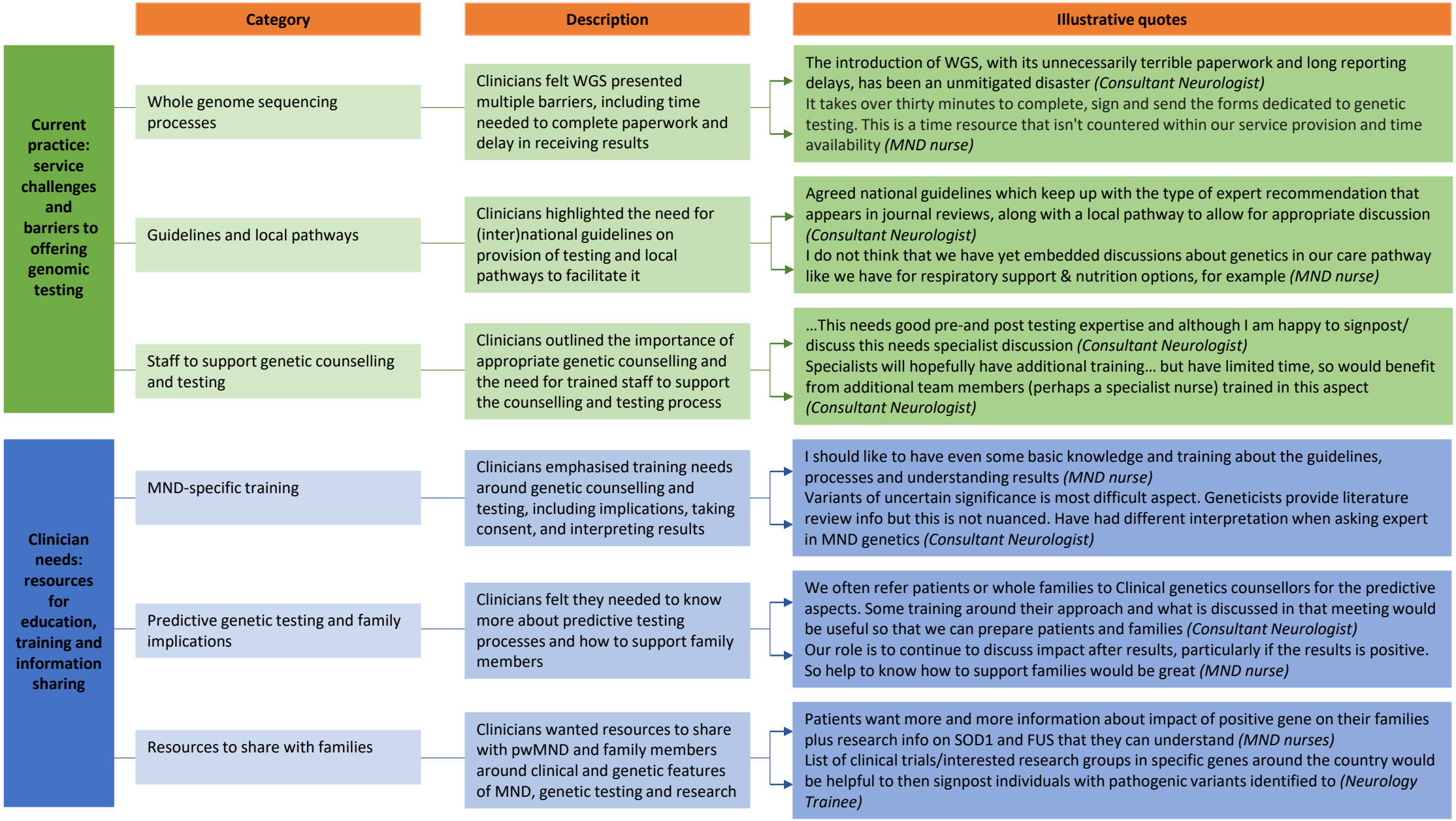
Category
Description
Illustrative quotes
Current practice: service challenges and barriers to offering genomic testing
Whole genome sequencing processes
Clinicians felt WGS presented multiple barriers, including time needed to complete paperwork and delay in receiving results
The introduction of WGS, with its unnecessarily terrible paperwork and long reporting delays, has been an unmitigated disaster (Consultant Neurologist)
It takes over thirty minutes to complete, sign and send the forms dedicated to genetic testing. This is a time resource that isn't countered within our service provision and time availability (MND nurse)
Guidelines and local pathways
Clinicians highlighted the need for (inter)national guidelines on provision of testing and local pathways to facilitate it
Agreed national guidelines which keep up with the type of expert recommendation that appears in journal reviews, along with a local pathway to allow for appropriate discussion (Consultant Neurologist)
I do not think that we have yet embedded discussions about genetics in our care pathway like we have for respiratory support & nutrition options, for example (MND nurse)
Staff to support genetic counselling and testing
Clinicians outlined the importance of appropriate genetic counselling and the need for trained staff to support the counselling and testing process
…This needs good pre-and post testing expertise and although I am happy to signpost/ discuss this needs specialist discussion (Consultant Neurologist)
Specialists will hopefully have additional training… but have limited time, so would benefit from additional team members (perhaps a specialist nurse) trained in this aspect (Consultant Neurologist)
Clinician needs: resources for education, training and information sharing
MND-specific training
Clinicians emphasised training needs around genetic counselling and testing, including implications, taking consent, and interpreting results
I should like to have even some basic knowledge and training about the guidelines, processes and understanding results (MND nurse)
Variants of uncertain significance is most difficult aspect. Geneticists provide literature review info but this is not nuanced. Have had different interpretation when asking expert in MND genetics (Consultant Neurologist)
Predictive genetic testing and family implications
Clinicians felt they needed to know more about predictive testing processes and how to support family members
We often refer patients or whole families to Clinical genetics counsellors for the predictive aspects. Some training around their approach and what is discussed in that meeting would be useful so that we can prepare patients and families (Consultant Neurologist)
Our role is to continue to discuss impact after results, particularly if the results is positive. So help to know how to support families would be great (MND nurse)
Resources to share with families
Clinicians wanted resources to share with pwMND and family members around clinical and genetic features of MND, genetic testing and research
Patients want more and more information about impact of positive gene on their families plus research info on SOD1 and FUS that they can understand (MND nurses)
List of clinical trials/interested research groups in specific genes around the country would be helpful to then signpost individuals with pathogenic variants identified to (Neurology Trainee)