Post-Meeting Notes: Q&A with Dr Sarah Gosling, PhD Physiotherapist – Fabry and Physical Activity

We were delighted to host Dr Sarah Gosling, physiotherapist and researcher, to discuss her work on Fabry disease and physical activity. The session offered practical, evidence-informed advice while recognising the very individual nature of Fabry symptoms and how these can impact exercise and movement. Below is a summary of the key themes.

1. Exercise during or after pain crises (acroparesthesias)

- Pain is highly individual – there is no single approach that works for everyone.
- Aim for a continuum of activity rather than an "all-or-nothing" approach, which can lead to frustration and a sense of failure.
- Reducing sedentary time – breaking up long periods of sitting may make symptoms more tolerable.
- Hands: gentle stretching and movement exercises.
- Stretching: hold for around 10 seconds; avoid pushing past a 6/10 on the pain scale.
- Feet: rolling, circles, and seated movements can be useful.
- Heat therapy: may help by dilating blood vessels and improving circulation.
- **Overall goal**: increase blood flow, supporting nerves and maintaining range of motion.

2. Managing heat intolerance and activity in hot conditions

- Many people with Fabry experience difficulties regulating temperature.
- Even small reductions in sedentary behaviour (for example, moving during TV advert breaks) can make a difference.
- Begin with seated exercises to build tolerance gradually.
- The psychological benefit of achievable movements should not be underestimated.

3. Fatigue management and pacing

- Plan activities in manageable chunks – remembering that "manageable" will look different for each individual.
- Prioritise lower-intensity or seated exercises when fatigued.
- Use pacing strategies: start gently and increase slowly.
- Incorporate breathing techniques to regulate heart rate and manage exertion.

4. Balance and fall prevention

- Neuropathy can reduce sensation in the feet, leading to instability.
- Work on lower limb stretches and movements to stimulate circulation and neural pathways.

- Train balance safely through replicated movements, resistance band work, or single-leg activities.
- Adapt sessions to avoid overheating, which may exacerbate symptoms.

5. Exercise with cardiac involvement

- Always seek consultant approval before making exercise changes.
- Physiotherapy programmes typically address:
    - Balance
    - Flexibility
    - Strength (including seated options)
    - Aerobic fitness (for example, walking)
- Patients are encouraged to advocate for themselves – keeping notes, setting goals, and highlighting gaps in current clinical guidance to their medical team.

6. Everyday stretches and joint comfort

- Regular, gentle stretching can help reduce stiffness and maintain mobility.

7. Physical activity and disease progression

- At present, no direct evidence shows physical activity prevents organ complications in Fabry.
- However, strong evidence from other conditions shows exercise supports cardiac, renal, and respiratory health.

8. Flare-ups after activity

- Pacing is essential – begin with seated, light exercises and increase slowly.
- Current research in Fabry is limited to those with moderate symptoms, for safety reasons. Over time, it is hoped that future studies will include people with more severe symptom profiles.

9. Adapted exercise programmes

- No formal Fabry-specific exercise programme currently exists.
- Tailoring to the individual is key. Options that may be helpful include:
    - Hydrotherapy: supportive environment, helps with circulation and neuropathy.
    - Pilates or Tai Chi: gentle strengthening and balance, available in group or online formats.

10. Tracking progress

- Progress is highly personal – define what it means for you.
- Possible measures include:
    - Step count and aerobic capacity

    - Weekly or daily activity logs
    - Symptom diaries
    - Smartwatch data (for example, heart rate, VO_2 max)
- Goals should be realistic, achievable, and flexible to account for symptom variation.

11. Mental health benefits

- Exercise supports mental wellbeing and quality of life through the release of endorphins.
- Activities outdoors or with a social element (for example, walking groups or classes) are especially valuable.

Ongoing research with Dr Gosling

Dr Gosling's current project is exploring a structured programme incorporating:

1. Individual consultations and goal-setting.
2. Regular progress check-ins.
3. A website with accessible, evidence-based resources.
4. Smartwatch integration to monitor activity.
5. Peer support groups for motivation and shared learning.

Breathing and heart rate management are also emerging as important techniques – helping individuals pace themselves and regulate exertion safely.

These insights are intended as general guidance. All exercise should be adapted to the individual, and medical advice should always be sought before making significant changes to activity levels.