# Fabry Disease

**Fabry disease is a rare genetic condition causing the buildup of a specific fat in cells. Beginning in childhood, this buildup causes signs and symptoms that affect many parts of the body, causing pain, skin lesions and rash, reduced sweating, corneal opacity, gastrointestinal issues, tinnitus, and hearing loss. Serious complications include kidney damage, heart attack, and stroke.**

## School absence patterns

- Absence pattern is typically intermittent and indefinite in length
- One day missed per fortnight for medical infusions (~30% of Australian Fabry patients qualify for LSDP-subsidised treatment due to end-organ damage symptoms)
- Additional days for flare-ups, medical appointments, recovery, fatigue

## Challenges for students

- Increased anxiety and mental health challenges
- Fatigue and pain reduce focus, performance, and physical participation
- Absences causing academic and social challenges
- Disruption to, and difficulty forming friendships
- Feeling invalidated by peers or teachers
- Pressure to explain or justify absences or limitations

## Challenges for families

- Challenges likely differ for patients with inherited (with other family affected) vs. de novo (not appeared in previous generations) Fabry mutations
- Managing emotional stress, advocacy, and privacy
- Adjusting routines for treatments
- Siblings (without Fabry) may feel anxious or overlooked
- Navigating stigma and community misunderstanding

## What schools should know

- Fabry is an invisible disease; students may appear fine but still experience pain or fatigue
- Symptoms vary widely and fluctuate between normalcy and flare-ups
- School absence reflects health challenges, not lack of commitment
- Empathy and understanding reduce stigma and foster inclusion
- Respect student and family privacy; disclosure may be limited
- Work with families to develop Individual Learning Plans (ILPs) with flexible deadlines for assignments and assessments; adjustments for physical activities and excursions; and flexible attendance policies for medical needs
- Assign a trusted staff member as a point of contact
- Prepare contingency plans for absences and catching up on work
- Create an inclusive environment where students feel safe sharing their needs

MISSING SCHOOL

## Health condition

- Gastrointestinal issues (pain, nausea, bloating, vomiting)
- Neuropathic pain in extremities
- Skin lesions, rash
- Pain crises involving multiple severe symptoms
- Heart, kidneys, brain, nervous system, lungs and eyes can also be affected
- Fatigue and mental health issues

## Student population

No specific data available on school-aged children

Estimated at **1 to 5 in 10,000 increasing to 1 in 3,000** including late onset variants

Approximately **500 students are currently diagnosed** in Australia

**Often undiagnosed or misdiagnosed** due to rarity and variability

**Only a quarter of diagnoses are made in childhood**

*"I felt a lot of pressure every time I needed to miss school, for academic and social reasons... Quite often I would go to school—having had a really terrible night or feeling pain—because I felt I had missed too much ... [so it] affects a lot of different elements of wellbeing and mental health."*

FABRY AUSTRALIA

**Fabry Australia**

A patient-run non-profit organisation that has been uniting and supporting the Australian Fabry community since 1994

**www.fabry.com.au**