Fabry Disease: An Underdiagnosed Disorder[1,2]

In Fabry disease, delay between symptom onset and diagnosis may be 10 years.[1,2]

FABRY FAMILY TREE SCREENING

Diagnosis of Fabry disease

The average age of diagnosis of Fabry disease is approximately 24 years for males and 31 years for females.[3] Due to symptoms easily mistaken for symptoms of other, more common conditions and heterogenous, multi-system manifestations, diagnostic delays have been observed in both genders.[4,5]

Clinical manifestations of Fabry disease

Childhood and Adolescence: Symptoms include neuropathic pain, acroparesthesia, angiokeratoma, gastrointestinal symptoms, reduced sweating and exercise intolerance, microalbuminuria, and hearing loss / tinnitus.[1,6,7]

Adulthood: Fabry disease may progress to chronic kidney disease, neurological complications, cerebrovascular disease, cardiac arrhythmias, hypertrophic cardiomyopathy, and heart failure, which could reduce life expectancy by approximately 5-14 years in women and 16 years in men.[8,9]

Ocular manifestations of Fabry disease

Ocular manifestations include cornea verticillata, conjunctival vessel tortuosity, retinal vessel tortuosity, and Fabry cataract.[10]

Early diagnosis and disease management may help improve symptoms of Fabry disease and reduce organ damage, morbidity, and premature mortality.[11]

A multidisciplinary approach involving cardiologists, nephrologists, psychologists, geneticists, and allied professionals is essential to provide genetic counseling, conduct family screening, manage the clinical and psychosocial needs of patients, and ensure appropriate disease management.[5,12]

Understanding the inheritance patterns of Fabry disease can help diagnose patients at an earlier age.[1]

FD: Fabry disease.

Family screening helps in the early identification of Fabry patients and indicates relatives who should consider being screened.[1]

Family screening can identify, on average, 5 family members with Fabry disease following the diagnosis of an index patient.[13]

Family trees based on family history can be a helpful tool for patients to understand who in the family may be at risk.[1]

PEDIGREE ANALYSIS

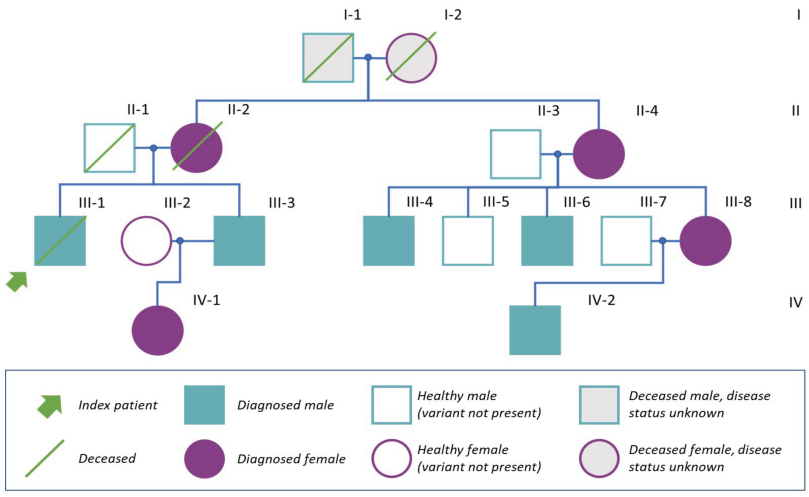
A geneticist or genetic counselor can support with pedigree analysis and help identify at-risk family members due to X-linked inheritance.[1,12]

A diagnosis can help family members understand previously unexplained symptoms

Risk status across family trees

Of the 16 family members shown, 7 living family members are affected by Fabry disease

Four generation pedigree of a family impacted by Fabry Disease



The use of family tree screening can benefit Fabry patients, educate patients on inheritance patterns, and potentially impact the lives of patients and their families.[1,5,12]

FD: Fabry disease.

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