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MISDIAGNOSIS OF ANDERSON FABRY DISEASE: DATA FROM FOS - THE FABRY OUTCOME SURVEY

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Fabry outcome survey (FOS) is a European database of Anderson-Fabry disease (FD) and the effect of enzyme replacement therapy (ERT) with agalsidase alfa (Replagal). 336 patients aged from 9 months to 71.7 years are currently enrolled. FD is an X-linked lysosomal storage disease, due to alpha-galactosidase deficiency, leading to reduced quality of life and life expectancy. It is commonly diagnosed in adulthood with progressive renal insufficiency, hypertrophic cardiomyopathy and cerebrovascular accidents.

FD affects both males and females similarly. A recent analysis of FOS data on 35 children (<18 y) has demonstrated that young patients are also frequently affected with severe signs and symptoms. Common clinical manifestations in childhood were cornea verticillata, angiokeratoma, acroparaesthesia and gastrointestinal problems. FD is often misdiagnosed. We report FOS data on the frequency of mistaken diagnosis of this slowly progressive and fatal disease. Misdiagnoses were reported in 67 (24.5%) of 274 patients for whom information on previous diagnoses was available. The most frequent misdiagnoses were rheumatological disease (13%), neuropsychological disease (3.3%), fibromyalgia (1.8%), Osler Weber Rendu (1.1%), dermatomyositis (1.1%) and erythromyalgia (1.1%). 21 of the 67 patients were seen by several specialists and received 2–4 different diagnoses, before FD was finally confirmed. The FOS database has clearly demonstrated the difficulty in diagnosis and lack of clinical awareness of signs and symptoms of FD, which often present in early childhood. A very high index of suspicion is needed amongst clinicians to make this diagnosis early, as there is clear evidence of beneficial effects of ERT.