Mitochondrial diseases are considered among the most common genetic metabolic disorders. A special group consists of lysosomal storage diseases, one form of which is Fabry disease (FD). This is a progressive orphan hereditary metabolic disorder linked to the X chromosome, with a mutation occurring in the GLA gene encoding the enzyme α-galactosidase A, resulting in decreased or absent activity of this enzyme, leading to abnormal accumulation of glycophospholipids and metabolic disturbance. Objective of this review article is to examine current literature on the pathogenesis, factors playing an important role in the disease pathogenesis, diagnostic algorithms and treatment of FD.

Materials and methods. This review article used works by Russian and foreign authors published on internet platforms and in print over the past 10 years.

Results. The review article discusses current literature on clinical manifestations, factors important in disease diagnosis, modern aspects of diagnosis and principles of FD treatment. Recent studies have also added information on the epidemiology of FD.

Conclusion. Diagnosis and treatment of FD have evolved significantly in recent years due to improved patient selection algorithms, progress in laboratory and instrumental diagnostics of the pathology, and increased accessibility of these methods for timely identification of clinical cases. However, a comprehensive understanding of the etiology, pathogenesis and consequently the clinical manifestations of the disease has still not been achieved, leading to late diagnosis and lack of timely pathogenetic therapy. Therefore, further research is needed to elucidate the factors involved in early diagnosis of the disease in patients with FD.