CASE REPORT: FIRST DOCUMENTED CASE OF FABRY DISEASE IN KAZAKHSTAN

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Introduction: Fabry disease (FD) is a rare, multisystem, X-linked lysosomal storage disease, resulting from a deficiency of the enzyme A-galactosidase A (A -Gal A). This results in the widespread lysosomal accumulation of the glycosphingolipid substrates of A -Gal A, particularly globotriaosylceramide (GL-3) in vascular endothelial cells.

Case report: We report a 38-year-old Asian male who presented with fifteen-years history of weakness, petechial-like angiokeratomas (umbilical, swimsuit region, and on extremities), painful acroparaesthesias (tingling, numbness, and stiffness of phalanges — particularly during a cold season) and valvular disease (mitral valve deficiency).The disease manifested in patient's adolescence, when he had painful feeling of tingling and numbness in his fingers. In his 20's he developed angiokeratomas around genitals and umbilicus which spread on the back, chest and the inner parts of upper and lower extremities. At the same time period patient experienced joint pains, non-specific pain and difficulties in breathing during physical exercises, and did not perspire in hot weather. Combined with non-specific cardiac symptoms, this led to a diagnosis of FD at the age of 36 years which was assigned in our clinic. Investigation revealed a c/[313A>T] (p.[R105*] mutation in the a-GAL gene and an abnormally low leukocyte activity of the a-GAL enzyme (0.1 μmol/l/h). Having confirmed diagnosis he referred to our hospital for enzyme replacement therapy (ERT).

During dermatologic examination we revealed diffuse red small skin lesions in gluteal, lower abdominal, lower back and inguinal regions. He had similar lesions around umbilicus; less numerous lesions were also present in the armpits and on the inner aspects of the thighs and shoulders. Appearances of these lesions were interpreted as angiokeratomas. Routine hematological tests were normal including the coagulation profile. Renal function was normal with estimated GFR using CKD-EPI 114.6 ml/min/1.73.m^2. His creatinine was at the level of 70 – 76 mmol/L, and hadn't increased.Electrocardiography didn't reveal any abnormalities at all. In other hand echocardiography disclosed normal systolic activity and mitral valve deficiency type 2. These findings were consistent with the diagnosis of FD. To prevent further development of FD and its complications including CKD we decided to start ERT. Fabrazyme was used to patient's main treatment. According to his body weight (70 kg) patient received 70 mg of Fabrazyme infusion in our clinic firstly, and then he was suggested to continue the treatment after 2 weeks.The patient was discharged and we will follow him up in our clinic every six months.

Discussion: The patient reportedhere was the first case diagnosed in Kazakhstan. Subsequent family studies revealed that his mother was acarrier of this X-linked recessive disorder, and the disease was diagnosed in 2 of his daughters.

Despite the fact that FD is one of the orphan diseases early diagnosis is important. As it being said FD have a lot of signs and symptoms as well as more severe complications. Therefore it is necessary to start ERT early. ERT has been shown to clear the accumulated GL-3 in the blood vessels as well as in the cells of the heart, kidney, and skin. Reported patient didn't have renal impairment symptoms (proteinuria, microscopic hematuria, and progressive CKD). And we suppose that ERT will help to prevent patient from CKD, which leads to ESRD.