Uloga medicinske sestre u otkrivanju i praćenju Anderson-Fabryove bolesti

Role of the nurse in detection and monitoring of Anderson-Fabry disease

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Anderson-Fabryova bolest je nasljedni nedostatak enzima alfa-galaktozidaze, karakteriziran nakupljanjem glikosfingolipida u lizosomima.[1,2] Pacijenti s Fabryovom bolešću ne mogu katabolizirati membranski neutralne glikosfingolipide, posebno globotriazilceramide, koji se stoga akumuliraju uglavnom u srcu, koži, bubrezima, krvnim žilama i središnjem živčanom sustavu. Bolest je recesivna i spolno povezana, prenosi se s X kromosomom. Karakteristični simptomi i znakovi Fabryove bolesti su prisutnost angiokeratoma (pojava vaskularnih kožnih lezija), akroparestezije (periodične bolne krize u ekstremitetima), hipohidroza (nemogućnost znojenja) i karakteristično zamagljivanje rožnice. Bolest počinje pri rođenju, tijekom djetinjstva obično je bez kliničke važnosti, organska oštećenja postaju očita tijekom četvrtog desetljeća života kod muškaraca i tijekom petog desetljeća kod žena.

Srčane manifestacije bolesti su rezultat nakupljenih globotriazilceramida u kardiomiocitima, stanicama i ventilacijskim sustavima, čija je glavna posljedica hipertrofična kardiomiopatija lijevog ili oba ventrikula. U naprednom stupnju bolesti, promjene srčanog mišića mogu dovesti do srčanog udara, kardiomiopatije i poremećaja provođenja. Ehokardiografija je izvrsna neinvazivna dijagnostička metoda za otkrivanje Fabryove bolesti (hipertrofija lijeve klijetke, stanje srčanih zalistaka i dimenzija srčanih komora).

Koordinacija liječnika i medicinskih sestara obavezna je za optimalno ehokardiografsko ispitivanje, detekciju i daljnji nastup bolesti. U bolesnika sa sumnjom na Fabryovu bolest medicinska sestra po nalogu liječnika uzima uzorke krvi kapilarno ili intravenozno. Nakon sušenja, uzorci se šalju na analizu enzimske aktivnosti i genetsko ispitivanje. Nakon dijagnosticirane i potvrđene Fabryove bolesti bolesnik svaka dva tjedna dolazi na terapije. Kontrola enzima se provodi svaka tri mjeseca kako bi se pratio učinak terapije. Praćenje bolesti provodi se kontrolom ultrazvuka, određivanju biomarkera (GL3, Lyso GL3) i antitijela tijekom primjene enzimatske nadomjesne terapije.

Anderson-Fabry disease is congenital deficiency in α-galactosidase A activity leading to intra-lysosomal accumulation of neutral glycosphingolipids.[1,2] Patients with this disease are unable to catalyze neutral glycosphingolipids, mainly globotriaosylceramide (Gb3), which re then accumulating in various organ systems, heart, skin, kidney, blood vessels, and central nervous system. The disease is an X-linked LSD inherited recessively. Characteristic symptoms and sings of Fabry disease include angiokeratoma (vascular skin lesions), acroparesthesiae (periodic painful crises in limbs), hypohidrosis (inability to sweat) and characteristic clouding of cornea. Disease occurs at birth, during childhood is usually of no clinical relevance, with organ defects obvious during fourth decade of life in men and fifth decade in women.

Cardiac manifestations of the disease are result of s associated with Gb3 accumulation in all cellular components of the heart, including cardiomyocytes, conduction system cells, valvular fibroblasts, endothelial cells and vascular smooth muscle cells, resulting in hypertrophic cardiomyopathy of the left or both ventricles. In advanced stage of the disease, changes in heart muscle can lead to the heart attack, cardiomyopathy and conductive disorders. Echocardiography is an excellent non-invasive diagnostic tool for diagnosis of Fabry disease (left increased left ventricular wall thickness, valvular changes, cardiac chambers quantitation).

Cooperation of physicians and nurses is imperative for optimal echocardiographic imaging, detection and monitoring of the disease. In patients with clinical suspicion of Fabry disease, nurse performs venepunction or take blood capillary. After drying, samples are sent for analysis of enzymatic activity and genetic testing. After diagnosis of Fabry disease, patient comes to therapy every two weeks. Enzyme control is performed every three months in order to monitor the effect of therapy. Monitoring of the disease includes ultrasonography, biomarkers (GL3, Lyso GL3) and antibodies during the application of enzyme substitution therapy.

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