UNIVERSIDAD SIMÓN BOLÍVAR | INSTITUTO DE POSGRADOS

REPORTE DE CASO: FAMILIA QUE PADECE ENFERMEDAD DE FABRY. ANÁLISIS DE SU EXPRESIÓN CLÍNICA

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RESUMEN

La Enfermedad de Fabry forma parte de las enfermedades ligadas a errores innatos del metabolismo, del subgrupo de enfermedades lisosomales; y se caracteriza por la deficiencia de la actividad de la enzima lisosomal llamada a-galactosidasa A (a-Gal A), enzima que interviene en el catabolismo de los glicoesfingolípidos. El defecto enzimático lleva a la acumulación de Globotriaosilceramida (GL-3) y de otros glicoesfingolípidos.

Mediante la realización de una encuesta familiar se evaluó grado de autoconocimiento de los otros miembros de la familia que padece FD. Valorando de esta manera logramos estimar su calidad de vida mediante, estrato socioeconómico, nivel de educación, edad, género, profesiones. Además se presentan dos casos clínicos de pacientes que cursan con FD.

Objetivos:
General
Analizar la expresión clínica y evaluación del conocimiento general sobre FD, pertenecientes a una misma familia de Armenia, Quindío.

Específicos
- Evaluar el fenotipo de los miembros de esta familia que ya sean diagnosticados con la FD.
- Determinar los factores de riesgo y complicaciones de la FD.
- Aplicar una encuesta al total de miembros afectados por FD de la misma familia que nos permita identificar sus características socioeconómicas.

- Identificar el perfil sociodemográfico y clínico de los pacientes, así como su calidad de vida.
- Establecer el grado de afectación de la enfermedad y relacionar con el nivel académico de los miembros que padecen esta patología.
- Evaluar el nivel de conocimiento de los pacientes con FD sobre su enfermedad y relacionarlo con su calidad de vida.

Materiales y Métodos:

La presente investigación es de tipo observacional descriptivo, exploratorio, con un enfoque cualitativo, de finalidad básica aplicada, con ubicación horizontal en el tiempo actual y de alcance exploratorio correlacional se realizará el reporte de dos casos de una misma familia que padece la FD. El diseño de la investigación fue de tipo no experimental: correlacional- transversal ya que no se manipuló ni se sometió a prueba las variables de estudio.

La población del estudio corresponde a una familia de 23 miembros, de los cuales hay 13 miembros diagnosticados con la FD, y 2 de ellos portadores asintomáticos.

La muestra corresponde a dos de los casos de una familia con pacientes confirmados genéticamente y enzimáticamente.

Además se realizó una encuesta familiar para determinar el grado de conocimiento de los pacientes de su propia enfermedad, el grado de afectación de la enfermedad en su diario vivir y su nivel socio económico.

Resultados:

El 35.2% son pacientes diagnosticados con FD, 64% son no portadores de la enfermedad y el 8.8% de los familiares fueron excluidos del estudio por vivir en otro país (España). Solo el 16% de pacientes con FD de esta familia decidieron participar en el caso clínico, el 84% restante decidió no participar por diferentes razones personales.

La muestra del estudio reveló un perfil sociodemográfico caracterizado por 9 individuos predominantemente masculinos (55.5%) y jóvenes, con una edad promedio de 40.4 años, educados con un título de educación superior (44.4%), solteros (66.66 %), casados (33.33%) y residentes en un área urbana (100 %).

Nuestros encuestados tenían un tiempo de diagnóstico promedio de 6 años, facto que todos los individuos hicieron el diagnóstico al mismo tiempo, y estaban principalmente acompañados por alguna especialidad médica (77.7 %), percibiendo sus síntomas como leves (55%). El tipo de tratamiento más común fue la terapia de reemplazo enzimático, registradas en 7 sujetos. La especialidad médica más consultada por los participantes fue la cardiología y la menos consultada fue la genética médica y la psicología. El 100% de la muestra dijo que no sentía la necesidad de obtener más información sobre la enfermedad.

El Sujeto 1, tiene diagnostico enzimático de FD desde el 26 de Junio del 2013. Su diagnóstico llego a raíz del diagnóstico de un primo lejano en la ciudad de Cali. Toda la familia fue citada para realización de pruebas y establecer si eran portadores o no de la FD. Masculino quien ha presentado múltiples hospitalizaciones por patologías secundarias a la FD. Repercusiones Neurológicas (2 Accidentes cerebrovasculares, ACV), repercusiones cardiovasculares (2 Infartos Agudos al Miocardio, IAM), Insuficiencia venosa y linfática periférica, Nefropatía por FD, Hipertensión Arterial.

El Sujeto 2, con diagnóstico genético de FD, quien se le ha realizado múltiples pruebas y valoraciones, se encuentra hasta el momento sin secuelas ni repercusiones de ningún tipo a raíz de la FD. Actualmente solo presenta un trastorno de la refracción, pero ello no es atribuible a la FD.

Conclusiones:

- El cuestionario puede ser una herramienta importante de monitoreo y apoyo conceptual para diseñar programas de educación para la salud basados en los déficits de conocimiento detectados, así como para realizar investigaciones científicas futuras. Sin embargo, consideramos que el instrumento que hemos elaborado no es perfecto ni está terminado.

- La FD es una patología altamente limitante cuando se presentan repercusiones como las del paciente Sujeto 1, ya que desde temprana edad comenzó a sentir toda la sintomatología periférica con dolores de etiología desconocida.

- Por la acumulación de esfingolípidos a nivel de todos los órganos del cuerpo, en especial Riñón, Corazón, y Cerebro se consideran comunes en estos pacientes los Accidente cerebrovasculares y los episodios de síndrome coronario.

- Paciente masculino quien al sufrir accidente laboral es llevado a osteosíntesis de huesos fracturados, cirugía fallida por rechazo al material de osteosíntesis, ello producido por la microangiopatía secundaria a FD.

- Paciente femenina Sujeto 2 quien se encuentra padeciendo una mutación de novo para diagnóstico de FD nunca antes reportada. Mutación no patológica.

- Entre las principales dificultades percibidas en el curso de este trabajo está la falta de un instrumento de medición previamente validado.

Palabras clave: Glicoesfingolípidos, Enfermedad Lisosomal, Alfagalactosidasa, Síndrome Fabry Anderson, Lisosoma, Enfermedad De Fabry.

ABSTRACT

Fabry's disease is part of the diseases linked to inborn errors of metabolism, of the subgroup of lysosomal diseases; and is characterized by a deficiency in the activity of the lysosomal enzyme called a-galactosidase A (a-Gal A), an enzyme that is involved in the catabolism of glycosphingolipids. The enzyme defect leads to the accumulation of Globotriaosylceramide (GL-3) and other glycosphingolipids.

By conducting a family survey, the degree of self-knowledge of the other family members suffering from FD was evaluated. In this way, we estimate their quality of life through socioeconomic status, level of education, age, gender, professions. In addition, there are two clinical cases of patients with FD.

Goals:

General

Analyze the clinical expression and evaluation of general knowledge about FD, belonging to the same family in Armenia, Quindío.

Specific

• Evaluate the phenotype of members of this family who are already diagnosed with FD.
• Determine the risk factors and complications of FD.
• Apply a survey to the total members affected by FD of the same family that allows us to identify their socioeconomic characteristics.
• Identify the sociodemographic and clinical profile of the patients, as well as their quality of life.
• Establish the degree of disease involvement and relate to the academic level of the members who suffer from this pathology.
• Evaluate the level of knowledge of patients with FD about their disease and relate it to their quality of life.

Materials and methods:

The present investigation is descriptive, exploratory, with a qualitative approach, with a basic purpose applied, with horizontal location in the current time and with a correlational exploratory scope, the report of two cases of the same family that suffers from FD will be carried out. The research design was non-experimental: correlational-cross-sectional since the study variables were not manipulated or tested.

The study population corresponds to a family of 23 members, of which there are 13 members diagnosed with FD, and 2 of them asymptomatic carriers.

The sample corresponds to two of the cases of a family with genetically and enzymatically confirmed patients.

In addition, a family survey was conducted to determine the degree of knowledge of patients of their own disease, the degree of disease involvement in their daily lives and their socio-economic level.

Results:

35.2% are patients diagnosed with FD, 64% are not carriers of the disease and 8.8% of family members were excluded from the study because they lived in another country (Spain). Only 16% of patients with FD of this family decided to participate in the clinical case, the remaining 84% decided not to participate for different personal reasons.

The study sample revealed a sociodemographic profile characterized by 9 predominantly male (55.5%) and young individuals, with an average age of 40.4 years, educated with a higher education degree (44.4%), single (66.66%), married (33.33 %) and residents in an urban area (100%).

Our respondents had an average diagnosis time of 6 years, in fact all the individuals made the diagnosis at the same time, and were mainly accompanied by some medical specialty (77.7%), perceiving their symptoms as mild (55%). The most common type of treatment was enzyme replacement therapy, recorded in 7 subjects. The medical specialty most consulted by the participants was cardiology and the least consulted was medical genetics and psychology. 100% of the sample said they did not feel the need to obtain more information about the disease.

Subject 1 has an enzymatic diagnosis of FD since June 26, 2013. His diagnosis came as a result of the diagnosis of a distant cousin in the city of Cali. The whole family was summoned to carry out tests and establish whether or not they were carriers of the FD. Male who has presented multiple hospitalizations for pathologies secondary to FD. Neurological Implications (2 Stroke, CVA), cardiovascular repercussions (2 Acute Myocardial Infarction, AMI), Peripheral venous and lymphatic insufficiency, FD Nephropathy, Arterial Hypertension.

Subject 2, with a genetic diagnosis of FD, who has undergone multiple tests and assessments, has so far been without sequelae or repercussions of any kind due to FD. Currently, there is only one refractive disorder, but this is not attributable to FD.

Conclusions:

• The questionnaire can be an important monitoring and conceptual support tool to design health education programs based on the knowledge deficits detected, as well as to carry out future scientific research. However, we consider that the instrument we have developed is not perfect or finished.

• FD is a highly limiting pathology when there are repercussions such as those of the patient Subject 1, since from an early age he began to feel all the peripheral symptoms with pains of unknown etiology.

• Due to the accumulation of sphingolipids at the level of all the organs of the body, especially Kidney, Heart, and Brain, cerebrovascular accidents and episodes of coronary syndrome are considered common in these patients.

• Male patient who, when suffering an occupational accident, is taken to osteosynthesis of fractured bones, surgery failed due to rejection of the osteosynthesis material, this produced by microangiopathy secondary to FD.

• Female patient Subject 2 who is suffering from a de novo mutation for diagnosis of FD never previously reported. Non-pathological mutation

• Among the main difficulties perceived in the course of this work is the lack of a previously validated measuring instrument

KeyWords: Fabry, Glycosphingolipids, Lysosomal Disease, Alphagalactosidase, Fabry Anderson Syndrome, Lysosome, Fabry Disease.

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