Abstract 1237

LABORATORY DIAGNOSIS OF SEVERE HYPERTRIGLYCERIDAEMIA. CASES FROM THE DYSLIPIDAEMIA REGRISTY OF THE SPANISH ATHEROSCLEROSIS SOCIETY

Type: Abstract Submission EAS

Topic: ASA03 - DYSLIPIDEMIA AND RISK FACTORS / ASA03-05 Inherited dyslipidemias

Authors: M.J. Ariza[1], J. Rioja[1], O. Muñiz-Grijalvo[2], T. ARROBAS VELILLA[3], E. Ortega[4,5], D. Zambón[4,5], M. Domenech[4,5], L. A. Álvarez-Sala[6], J. Delgado-Lista[7], A. González-Estrada[2], V.A. Seidel[8], F. J. Fuentes[7], A. Camacho[9], M. J. Romero[9], M.Á. Sánchez-Chaparro[1,10], P. VALDIVIELSO FELICES[1,10]; [1]University of Málaga, Lípids and Atherosclerosis Laboratory. CIMES., Málaga, Spain, [2]University Hospital Virgen del Rocío, UCERV–UCAMI, Internal Medicine Department, Sevilla, Spain, [3]servicio andaluz de Salud, Clinical Biochemistry. Hospital Universitario Virgen Macarena de Sevilla, Sevilla, Spain, [4]Institut d'Investigacions Biomèdiques August Pi I Sunyer, IDIBAPS, Barcelona, Spain, [5]Hospital Clínic Barcelona, Endocrinology and Nutrition, Barcelona, Spain, [6]University Hospital Gregorio Marañón, IiSGM, Lipids Unit, Internal Medicine, Madrid, Spain, [7]Reina Sofia University Hospital. CIBER Fisiopatología de la Obesidad y Nutrición., Lipids and Atherosclerosis Unit, Internal Medicine Unit, Cordoba, Spain, Spain, [8]University Hospital Gregorio Marañón, Clinical genetics. Pediatric service, Madrid, Spain, [9]Hospital Infanta Elena, Vascular Risk Unit, Huelva, Spain, [10]University Hospital Virgen de la Victoria, Internal Medicine Unit, Málaga, Spain

Background and Aims

Severe hypertriglyceridaemia (sHTG) increases the risk of cardiovascular disease and acute pancreatitis episodes. Patients with sHTG fit mainly into two clinical entities: Familial or Multifactorial Chylomicronemia Syndromes (FCS and MCS, respectively). FCS and MCS exhibit clinical differences but also separate genetic and biochemical characteristics that can be assessed in the laboratory. The aim of this work has been to implement a laboratory workflow to help diagnose sHTG patients with either FCS or MCS.

Methods

Patients with two fasting triglycerides >1000mg/dL determinations were sequenced with a capture probe panel of 24 triglycerides-related genes using massive parallel sequencing (n=200). Two-step sequential ultracentrifugation was performed (n= 159) to diagnose Type I hyperlipoproteinemia (HLP I) and post heparin lipoprotein lipase activity was measured to discard or confirm its deficiency (n=60).

Results

Most patients had MCS as they: (i) did not exhibit HLPI and/or (ii) their genetic profile was not compatible with FCS and (iii) were not deficient in LPL activity. FCS cases were identified as they had: (i) HLPI, and/or (ii) biallelic pathogenic variants in *LPL* (n=5), GPIHBP1 (n=3), or *LMF1* (n=2) genes and/or (iii) LPL activity deficiency. We identified 4 FCS patients with HLPI, biallelic pathogenic variants in *APOA5* but a rescued LPL activity. An additional study of Apo-AV functionality was designed to confirm the FCS diagnosis in these cases.

Conclusions

Laboratory studies, in patients with severe hypertriglyceridaemia, provide with information of clinical utility to distinguish between Familial and Multifactorial Chylomicronemia Syndromes.

Print