P-114 - TWO GENETIC VARIANTS RELATED IN THE CLINICAL HETEROGENEITY OF A PEDIATRIC PATIENT

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\textbf{INTRODUCTION:} inborn errors of metabolism (IEM) are considered genetic disorders determined by blocking a metabolic step due to mutation of genes responsible for the operation of the same. The identification of genetic variants that affect gene expression and are related to pathological characteristics in diseases allow us to perform a personalized medicine and impact in the detection, treatment, prognosis, follow-up and Prevention through appropriate and assertive genetic counseling. 

\textbf{OBJECTIVE:} To determine the etiology of the clinical heterogeneity of a pediatric patient through complete exome sequencing

\textbf{METHODS:} a 14-year-old male patient diagnosed with multiple congenital malformations, severe psychomotor retardation, intellectual disability, severe muscular atrophy, multiple osteoarticular malformations, globular abdomen, joint hypermobility and bone deformations. Initial biochemical and normal imaging studies were performed. Given the complexity of the clinical manifestations of the patient, complete exome sequencing was performed in search of gene alterations. 

\textbf{RESULTS:} A pathogenic variant was identified in heterozygosis in the FGF23 gene, this result being compatible with the diagnosis of autosomal dominant hypophosphatemic rickets (ADHR). Likewise, a variant of uncertain significance was identified in homozygosis in the ABCD4 gene. This result is possibly compatible with the diagnosis of methylmalonic acidemia with homocystinuria type cbI\textsubscript{J}, which is transmitted in an autosomal recessive manner. The diagnosis of ADHR is an IEM characterized by renal loss of phosphate, hypophosphatemia and inappropriate levels of 1,25-dihydroxyvitamin D3. Patients often suffer from bone pain, rickets and abscesses. Likewise, methylmalonic acidemia with homocystinuria type cbI\textsubscript{J} is also an IEM, however, in this case there is a deficiency of vitamin B12 (cobalamin) which leads to conditions for megaloblastic anemia, lethargy, growth and development delay, intellectual deficit , and convulsions. 

\textbf{CONCLUSION:} The performance of genetic tests supports and guides the making of medical decisions based on the patient's clinical history, through the identification of the causes of complexity, due to the presence of different genetic variants and the heterogeneity of their expression, which leads to the practice of a precision and predictive medicine, which allows an early diagnosis, directed and personalized treatment, monitoring and appropriate genetic counseling.