Multicentric study. Population screening of hypophosphatasia. A metabolopathy to consider

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INTRODUCTION:

Knowing the profile of the different congenital errors of the metabolism (ECM), with its clinical and biochemical phenotype valued on the basis of its natural history, is the first to improve the diagnosis and therefore the prognosis of these diseases
Hypophosphatasia is a congenital disease, characterized by a defect in bone and dental mineralization, secondary to a deficiency in the biosynthesis of the non-specific tissue isoenzyme of bone, liver and kidney alkaline phosphatase (TNSALP).
Clinical phenotype varies with age and its clinical expression is sometimes very latent.
There is a small but significant number of pediatric patients NOT diagnosed with hypophosphatasia. The values of low phosphatases may go unnoticed in routine clinical practice. These children could benefit from a possible treatment that prevents the progression of symptoms.

OBJETIVE:

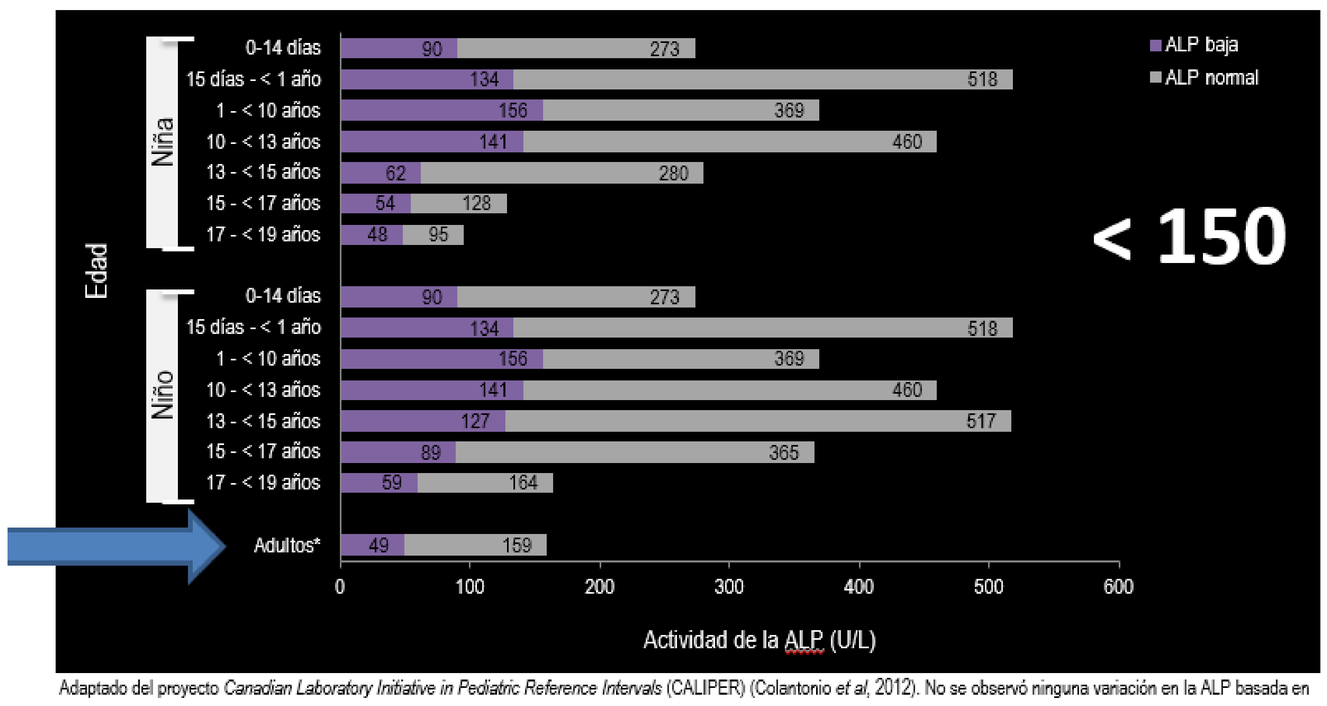
Identify those patients who gave values below normal (HYPOPHOSPHATASE) and assess whether these values can be biomarkers of rare disease. It is the first population screening study of these characteristics carried out in the region and at the national level.

Establish cut-off points adjusted for age and sex for alkaline phosphatase levels in relation to rare metabolic disease.
To evaluate the utility of retrospective studies in the diagnosis of rare diseases.

MATERIAL & METHODS:

Retrospective search during a 12-month period of the very low alkaline phosphatase values at pediatric age in the databases of the laboratory analyzes carried out at the Central Laboratory of the hospitals participating in the study. It will be verified also present low levels in the analytical carried out in other dates to discard, since the levels of alkaline phosphatases. Detected the case of very low values of alkaline phosphatases, will contact the pediatrician of children to know if they have any clinical data that is indicative of this disease

Normal values (IU / l) were considered - Clinical Chemistry Study 58: 5 (2012)



Adaptado del proyecto *Canadian Laboratory Initiative in Pediatric Reference Intervals* (CALIPER) (Colantonio *et al*, 2012). No se observó ninguna variación en la ALP basada en

RESULTADOS

Centro	Num de casos / total	Potenciales	Estudiados
HU Donostia	180	33	0
HU Araba	404		8
HU Cruces	927 / 15406	67	6
HU Basurto	1969	2	0
Total			14

RESULTS:

Population size: 16,555
Expected proportion: 0.0005%
Confidence level: 95.0%
Cases studied with initial PA + 3.480, confirmed potential cases 102.

Sent to genetic study and CCEE of Metabolopathies 14.
Exclusion data: Anorexia nervosa, Oncology / hematology, Neuromuscular disease
Psychiatric pathology, Obesity, Traumatology, Complex cardiology Endocrinology (poly ovary, growth retardation)

Cases studied (14): 8 cases associated with scoliosis (traumatology), 1 case associated with precocious puberty, 1 case of short stature, 1 case of liver disease and myasthenia, 1 case of teething problems

CONCLUSIONS:

Pending genetic confirmation, we present the first study of these characteristics in our territory
Low FA levels in many cases are transient
In patients with low AF the clinic should be evaluated, in order to select the possible cases.
PPH with little clinical phenotype may be more at the level of trauma.
IT alert could be assessed, in values lower than 20% to the limit of normality adjusted to sex and age