INTRODUCTION:
Glycine is a non-essential aminoacid, mainly an inhibitory neurotransmitter at specific receptors in the brain stem and medulla. Glycine encephalopathy, is an innate error of metabolism defined by a mutation of the GCS glycine cleavage enzymatic system. This enzyme defect results in the accumulation of large amounts of glycine. Excessive activation of N-methyl-D-aspartate receptors, results in neuronal and axonal damage, as well as altering neurogenesis. This condition has autosomal recessive inheritance from both parents carrying mutations in one of these genes, giving rise to a patient with both copies of the gene.

EPIDEMIOLOGY:
Described to date with an overall incidence of 1 in 76,000 children. Unfortunately in Colombia we do not have our own epidemiological data.

REFERENCES:
Clinical, biochemical characteristics and molecular studies of 38 patients with nonketotic hyperglycinemia in Colombia

MATERIALS AND METHODS:
Descriptive study that included all patients with glycine encephalopathy diagnosed at the Pablo Tobón Uribe Hospital (HPTU) in the city of Medellín Colombia between the period 2011 and 2021.
Clinical findings, biochemical, electroencephalographic and neuroimaging data in each child were described.
The qualitative variables included were described as frequencies, and of the quantitative variables those with a normal distribution were described as means with their respective standard deviations, finally, those that did not have a normal distribution were described as medians with their interquartile ranges.

RESULTS: 38 patients were obtained.
31% described paternal consanguinity and 42% of them had a history of early infant deaths as part of the family history.
The onset of symptoms was the first 7 days of life in 91.9% of the cases.

Results of glycine in CSF, plasma and relation of both in children with NKH Colombia

<table>
<thead>
<tr>
<th></th>
<th>Plasma nmol/ml</th>
<th>CSF nnmol/ml</th>
<th>CSF/plasma</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mean</td>
<td>906</td>
<td>167</td>
<td>0.21</td>
</tr>
<tr>
<td>Minimum</td>
<td>184</td>
<td>28</td>
<td>0.02</td>
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<tr>
<td>Maximum</td>
<td>2622</td>
<td>793</td>
<td>0.8</td>
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<tr>
<td>No data</td>
<td>3</td>
<td>9</td>
<td>9</td>
</tr>
<tr>
<td>Total n</td>
<td>38</td>
<td>38</td>
<td>38</td>
</tr>
</tbody>
</table>
Clinical, biochemical characteristics and molecular studies of 38 patients with nonketotic hyperglycinemia in Colombia

Findings in resonance and spectroscopy in children with NKH in Colombia

**DISCUSSION**

The present study exemplifies that for the Colombian population described, NKH is an important metabolic disease with a possible founder effect (same mutation GLDC c.2714T>G;(C2714T>G)* pVal905G) in this region, in part due to the high presence of consanguinity. Our presentations mostly correspond to severe phenotype, with early symptoms, biochemical, imaging and electroencephalographic findings that support the diagnosis. Without describing in the present study any patient with transient NKH. It is important to establishing protocols for diagnosis, multidisciplinary treatment, looking for expanding the knowledge of this disease.