INTRODUCTION: Homocysteinemia is defined as elevation of the homocysteine level in blood. Homocysteine is metabolized by transsulfuration and remethylation, cofactors are necessary in its metabolism. The transsulfuration to cysteine is catalyzed by cystathionine-beta-synthase and requires pyridoxal phosphate (B6) as a cofactor. Remethylation of homocysteine to methionine is catalyzed by methionine synthase or by betaine-homocysteine methyltransferase and vitamin B12 is the precursor of methylcobalamin, the cofactor for methionine synthase. In many of these conditions early treatment can prevent severe clinical compromise.

OBJECTIVES: To present a case series of 15 patients diagnosed with homocysteinemia between January 2015 and December 2018 in a tertiary hospital in Argentina and to describe the clinical biochemical and molecular findings.

MATERIALS AND METHODS: We performed a retrospective review of Medical records of all patients referred to the metabolic service at the Hospital Italiano of Buenos Aires, between 2015 and 2018. Patients were included who presented with elevated HCY.

RESULTS: A group of 15 patients was included (46% female), within a range at diagnosis of 1 months - 38 years (with a median of 6 months). The diagnosis were 2 (13.3%) CBS deficiency, 2 (13.3%) congenital malabsorption of B12, 4 (26,6%) Cobalamin metabolism defect (MTR, MTRR, and MMADHC), 1 (6.6%) pernicious anemia and 4 (26,6%) had B12 deficiency but no further studies were done. In 2 (13.3%) the condition was secondary to maternal disease. Most common clinical presentation was failure to thrive, anemia, hypotonia, seizures and encephalopathy. Treatment was initiated and changed the outcome in 15/15 patients. 10/15 had complete recovery.

DISCUSSION: Cobalamin metabolism disorders should be evaluate early in the diagnostic algorithm in any patient with suggestive clinical findings. Biochemical and molecular tools are available to determine the specific metabolic defect, and early treatment should be started as reversion of symptoms is frequent. We want to emphasize that total plasma homocysteine is a non expensive and usually available test that may change patients prognosis if used.