INTRODUCTION: Mucopolysaccharidosis IVA (MPS IVA; Morquio A) is an autosomal recessive lysosomal storage disease caused by deficiency of N-acetylgalactosamine-6-sulfate sulfatase (GALNS), leading to intracellular accumulation of keratan sulfate and chondroitin-6-sulfate. Clinical manifestations include short stature, hypermobile joints, and dysostosis multiplex, including atlantoaxial instability and cervical cord compression. They also present typical dysmorphisms, corneal opacities, hearing loss, myocardial insufficiency, valvular stenosis and obstruction of the respiratory tract. Cognitive function is minimally impaired. Clinical presentations reflect a spectrum of progression from classic (severe) to attenuated (mild) phenotype.

OBJECTIVE: To describe phenotype, clinical features and laboratory findings of patients diagnosed with MPS-IVA.

METHODS: Retrospective-prospective and descriptive study. Review of clinical records and laboratory findings of MPS-IVA patients who have been evaluated at the Pediatric Neurology Ward of San Borja Arriarán Clinical Hospital. Patients consent/assent was obtained. The study was approved by the Institutional Ethics Committee.

RESULTS: 12 patients (7 female), median age 13.79 years. Median age at clinical suspicion 1.99 years. Median age at diagnosis 5.54 years. Phenotype: classic 11 patients, attenuated 1 patient. All the patients with the classic phenotype showed 0-2% of normal GALNS activity in dried blood spot (DBS), while the patient with attenuated phenotype had GALNS activity less than 44% of the lowest control value. Neurological features: 10/12 patients presented developmental delay of motor skills and 8/12 severe cervical rachostenosis (2 complicated with myelopathy). Multisystemic illness: 9/12 sensorineural hearing loss, 8/12 alteration of cardiac and pulmonary function, 6/12 corneal opacities and 4/12 hepatomegaly. All patients have been treated with enzyme replacement therapy (median age at first dose 11.62 years, median total time on treatment 3.16 years), one discontinued due severe adverse effect.

CONCLUSIONS: almost all patients had a classic phenotype with only one patient with the attenuated form. This is consistent with the findings of the International Morquio Registry. A high phenotypic variability was observed, with the skeletal manifestations being the most consistent clinical feature with a wide range of non-skeletal features. This study provides an overview of the clinical manifestations and diagnosis of patients with MPS IVA in a chilean reference center.