San Filippo syndrome is part of a group of glycosaminoglycan metabolic disorders commonly named Muchopolysaccharidoses. It is an autosomal recessive inherited disorder affecting heparan sulfate metabolism via lysosomal enzymatic deficiency. Four different phenotypes have been described (A,B,C,D). This sub-classification can be done only by measurement of enzymatic catalytic activity on leucocytes or fibroblasts instead of clinically due to the variability in disease symptoms and affectations on osteoarticular and neurologic system among others. Here we present an approach to Type B San Filippo syndrome diagnosis through leucocyte enzymatic screening in a high-risk population.

**MATERIALS AND METHODS:** Analysis of leucocyte alpha-N-acetyl-glucosaminidase (NAGLU) was done by fluorometric method to assess cleavage of 4Methylumbelliferyl 2Acetamide 2Deoxy Alpha Dglucopyranoside used as a substrate in a buffer solution of citrate-phosphate 0.2M pH4.3. Screened population included 256 (0-20 years age) patients with MPS clinical suspicion and 450 normal controls were used to establish a normal range value.

**RESULTS AND CONCLUSIONS:** Registered range of activity for controls was 0.6-4.26 nmol/mgprotein/hour. From 256 patients screened, seven cases (2.7%) were detected (ages between 3-16 years). Activity range 0.0–0.24 nmol/mg protein/hour. More studies are needed to establish Colombian prevalence for this and the other 3 subtypes of the disease.