Genetic analysis of the SERPING1 gene in hereditary angioedema patients in Neiva, Colombia

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Background: Hereditary angioedema due to the deficiency of a serum protein called C1-inhibitor (C1-INH) has a frequency of 1:10000 to 1:50000 in the general population, able to induce swelling of sudden onset in the face, neck, arms, with a duration that may last up to five days. These symptoms begin in childhood, worsen around puberty and persist throughout life. C1-INH is a member of the serpin family of serine protease inhibitors whose gene SERPING1 is in chromosome 11q12- q31 with eight exons and seven introns.

Methods: 22 subjects were analyzed for mutation in the SERPING 1 gene by PCR (Centogene, Germany). C4, C1-INH concentrations and function were measure in all symptomatic individuals by nephelometry and ELISA, respectively.

Results: Three SERPING mutations were identified for the first time in Colombian subjects: c.1081 C > T, p.Gln361, c. 106_107 C >G pArg466Gly, and c.1396 C > G pArg466Gly.

Keywords: Hereditary angioedema; Genetic analysis