12º CONGRESSO BRASILEIRO DE ALERGIA E IMUNOLOGIA EM PEDIATRIA

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Trabalhos Científicos

Título: Caspase-8 Deficiency Manifesting As Adult-onset Multi-organ Granulomatous Disorder With Recurrent Infections.

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Resumo: CASP8 deficiency was described in a single family as a recessive disorder characterized by early-onset recurrent infections and apoptosis defects. Herein we present 2 siblings with CASP8 mutations and a different clinical phenotype. Pt1, female, was healthy until the age of 38, when she developed progressive dyspnea, pulmonary hypertension and interstitial lung disease. At the age of 42 she received a lung transplant, and died after 8m due to CNS nocardia infection. Pt2, male, was healthy until the age of 37 when he developed neurological symptoms caused by a granulomatous mass on Meckel's cave, recurrent pulmonary infections, and hepatosplenomegaly. All manifestations responded promptly to high dose steroids and mycophenolate mofetil. The family had their exomes sequenced and a recessive CASP8 c.1096C>T, p.Arg248Trp was identified in the affected siblings. These are the same mutations described on the original CASP8 deficient family, to which they are distantly related. In our patients, CASP8 deficiency presented as a late onset disease with end organ lymphocyte infiltrates, as described in elderly Casp8-/- mice.