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HUMAN INBORN ERRORS OF IMMUNITY: AN EXPANDING UNIVERSE

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DOUBLE TROUBLE- A DOCK8 AND CFI DEFICIENT INFANT PRESENTING WITH ACUTE NECROTIZING MENINGOENCEPHALITIS

EPOSTER VIEWING ONLY: ADAPTIVE IMMUNITY (T- AND B-CELLS)

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Background and Aims: Primary immunodeficiencies (PIDs) are rare disorders; it is also uncommon for two PIDs to coexist. DOCK8 deficiency is a combined immunodeficiency characterized by a predisposition to severe infections, eczema, allergies, autoimmunity, and malignancy. Complete Factor I deficiency is characterized by susceptibility to encapsulated bacterial infections.

Methods: Here we present a patient diagnosed as DOCK8 and CFI deficiency after acute necrotizing meningoencephalitis.

Results: A 45-day-old male patient was admitted to the pediatric intensive care unit (PICU) due to status epilepticus secondary to Streptococcus agalactia meningitis. A cranial MR was performed and revealed necrotizing meningoencephalitis and brain oedema. In spite of adequate treatment, his clinical status deteriorated on his 4th day of PICU stay. Increased intracranial pressure requiring external ventricular drainage was detected. New cerebrospinal fluid sampling revealed both S.agalactia and CMV positivity. On family history, we learned that parents were third degree cousins, and his elder brother was under IVIG treatment due to chronic mucocutaneous candidiasis and elevated IgE levels. Immunologic workup was performed and revealed low IgG, IgM and C3 levels and a reversed CD4/CD8 ratio. He also had CMV viremia and urinary tract infection with Candida albicans. Whole exome sequencing (WES) revealed a homozygous likely pathogenic variant [c.4507C>T (p. Gln1503*)] in DOCK8 gene and a homozygous likely pathogenic variant [c.262C>T (p. Gln88*)] in CFI gene. His brother had same mutation of DOCK8 gene but was a carrier for CFI gene.

Conclusions: Consanguinity increases the likelihood of PIDs. Therefore, family history and proper genetic investigations (particularly WES) are essential for diagnosis.

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Keywords: DOCK8 deficiency, necrotizing meningoencephalitis, CFI deficiency