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Clinical and Immunological Findings of a Child with Cell Division Cycle 42 mutation

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A male patient who is on 4,5 year of age, was admitted the neonatal intensive care unit because of anemia, thrombocytopenia, neutropenia and high acute phase reactans (APR) at the 20 day of age. Patient was diagnosed septicemia. At the 40 day of age, persistant fever, rash and hepatosplenomegaly developed. Patient was diagnosed Hemophagocytic lymphohistiocytosis, and treated intravenous immunoglobulin (IVIG) and steroid. An anemia, thrombocytopenia, neutropenia and high APR repeated at sixth month of age. Patient suffered from mucosal and intracranial bleading. Anemia and thrombocytopenia regressed, but neutropenia persisted in the following months. Pamidronate treatment was started for diagnosis of osteoporosis at the 21 month of age. Patient had hypotonia and mental, motor retardation. Fever with rash attacs started at 2 year of age, patient treated with anakinra for diagnosis of CAPS . There is no detected a mutation NLRP3 gene. An anemia without requirement of transfusion and neutropenia persisted on the following time. By the Whole exon sequensing, heterozygous missense variation
CDC42(LRG_1326t1:c556C>T;pArg186Cys) was detected. CDC42 is a member of the Rashomolog (Rho) GTPase family, which controls a range of cellular processes including adhesion, migration, polarity, cell cycle and proliferation. It acts as a key to control GTP and GDP conversions(1). NOARCH syndrome was newly described at 4 patients as neonatal onset pancytopenia, autoinflammation, rash and episodic HLH on 2019 (2).

References

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2. *Lam MT, Coppola S, Krumbach OHF, Prencipe G, Insalaco A, Cifaldi C,et al. A novel disorder involving dyshematopoiesis, inflammation, and HLH due to aberrant CDC42 function. J Exp Med. 2019 Oct 10. doi: 10.1084/jem.20190147.*

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