Abstract

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Identification of a New Candidate Gene for the Etiology of Periodic Fever Syndrome: CSF1R

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Background and aims: Periodic fever syndromes (PFS) are rare innate immunity disorders caused by single gene mutations triggering chronic/periodic systemic inflammation. Syndrome of Undifferentiated Recurrent Fever (SURF) definition includes all the cases lacking gene confirmation. Here we report the identification of a new candidate gene for PFS detected in a young patient.

Materials/patient and methods: Female, aged 15 years, our patient has been suffering from recurrent fever, arthralgias, pericarditis, peritonitis and oral aphtosis. The standard PFS genes panel was negative. Hence we performed a family whole exome sequencing (WES).

Results: We identified in our proband a CSF1R gene de novo transition (c.1735 C>T) causing the substitution p.Arg579Trp. CSF1R encodes the receptor of CSF1 and IL-34, two cytokines mediating macrophage production, differentiation and activation. CSF1R mutations are already described as risk factors for myeloid neoplasms.

Discussion: We detected a new PFS candidate gene that may be found in other SURF patients, leading to a better understanding of such syndromes and contributing to the improvement of their management. This should include for our patient a hematologic monitoring, since we also found another already known mutation in the same gene inherited from her mother, who was previously diagnosed a Hodgkin Lymphoma.