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An epigenetic mechanism is involved in the development of autoinflammatory diseases

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Researchers from the Chromatin and Disease group of the Bellvitge Biomedical Research Institute (IDIBELL), led by Dr. Esteban Ballestar, have identified for the first time epigenetic alterations in autoinflammatory diseases, particularly in cryopyrin-associated periodic syndromes (CAPS). The study has been published this week in the *Journal of Allergy and Clinical Immunology*, the most prestigious journal in the clinical immunology area.

Systemic autoinflammatory diseases comprise a set of rare diseases characterized by the presence of recurrent acute inflammatory episodes that result from the deregulation of control of the inflammatory process. These diseases occur as a result of mutations in different genes related to innate immune response and inflammation.

The three diseases that the term CAPS includes are Familial Cold Autoinflamatory Syndrome or FCAS, Muckle-Wells syndrome and Neonatal-Onset Multisystem Inflammatory Disease (NOMID) or Chronic Infantile Neurological Cutaneous Articular Syndrome (CINCA). All CAPS patients have mutations in the encoding gene cryopyrin, a key protein in the process of inflammation. Although the presence of mutations is common, not all patients present with the same clinical features or respond equally to drug treatment.

The study, which was mainly carried out by Drs Roser Vento-Tormo, Damiana Álvarez-Errico and Antonio García-Gómez, has not only shown that CAPS patients have alterations in the DNA methylation-

dependent control of inflammation genes in their monocytes, but also that the immunological treatments received by these patients are able to restore methylation levels to those observed in healthy individuals.

Since all CAPS patients neither have the same clinical features nor respond equally to treatment with immune drugs, the observed changes in the DNA methylation open up possibilities for the development of novel biomarkers with clinical uses, including a better characterization of patients as well as a follow-up in their response to treatments.

For this study, the close collaboration with clinicians involved in the diagnosis and treatment of these patients has been crucial. Specifically, the study has contributions from Drs. José Hernández-Rodríguez, Juan Ignacio Aróstegui, Jordi Yagüe and Manel Juan from Hospital Clínic de Barcelona, ??Dr. Segundo Buján from Hospital Vall d'Hebron, and Drs Maria Méndez and Maria Basagaña from Hospital Universitari Germans Trias i Pujol.

More information: Roser Vento-Tormo et al, DNA demethylation of inflammasome-associated genes is enhanced in patients with cryopyrin-associated periodic syndromes, *Journal of Allergy and Clinical Immunology* (2016). DOI: 10.1016/j.jaci.2016.05.016

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