ERRATUM

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The complete abstract is given below.
The publisher and organizers sincerely apologize for the errors and regret the inconvenience they may have caused.

SY1-3
NLRP3 somatic mosaicism on “NLRP3 mutation -negative” CINCA/NOMID patients

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CINCA syndrome/NOMID is an autoinflammatory disease whose triad consists of urticarial rash, arthritis/arthropathy, CNS symptoms due to chronic aseptic meningitis. NLRP3 gene has been reported as a responsible gene for the syndrome and the reported mutations are on the coding regions, especially missense mutations on exons 3, 4 and 6. The molecular mechanism underlying the CINCA/NOMID is believed to be dysregulated NLRP3 inflammasome activation by the NLRP3 mutations. Although the molecular mechanism is uncovered, it has been known that approximately 40% of CINCA/NOMID patients lack NLRP3 mutations on its coding regions, which suggests other genes mutations or NLRP3 non-coding regions mutations could be involved in these patients. We have recently reported NLRP3 somatic mosaicism occurs on some, but not all of the "NLRP3 mutation-negative" CINCA/NOMID patients (Saito, Blood 2008). We extended our study worldwide by collaborating with the researchers working on CINCA/NOMID (US, France, the Netherlands, Spain, Italy). Now we are analyzing the NLRP3 somatic mosaicism by subcloning-based strategy. I am going to talk about "NLRP3 mutation-negative" CINCA/NOMID and on-going study on these patients world-wide.