Singapore Immunology Network

A*STAR

SIgN SEMINAR

hosted by Dr Florent Ginhoux

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A novel missense mutation of a proteasome subunit, PSMB9, found in patients with autoinflammation and immunodeficiency

The proteasome is a large protein complex involved in degradation of unnecessary or useless proteins. Homozygous, compound heterozygous or digenic mutations of proteasome subunits cause a group of autoinflammatory diseases, which are now termed proteasome-associated autoinflammatory syndromes (PRAAS). However, it remains unknown how the mutation leads to autoinflammation, mainly because no mouse models are available. A de novo heterozygous missense mutation X in the proteasome subunit PSMB9 (encodes β1i) gene was found in two unrelated patients showing PRAAS-like manifestations and immunodeficiency. The mutation is novel and causes a substitution of an amino acid conserved among multiple species. We have generated the mutant mice carrying the Psmb9 X mutation by CRISPR/Cas9 technology and found that the heterozygous Psmb9 X mutant mice showed multiple defects in both innate and adaptive immunity. Our present findings should indicate a novel category of autoinflammatory diseases, distinct from PRAAS, as what we might call proteasome-associated autoinflammation and immunodeficiency disease.

8 January 2020 (Wednesday)
11:00am – 12:00pm
SIgN Seminar Room, Immunos Level 4

Seminar is open for all to attend.

Registration is not required.

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