Dokumenttyp: journal article

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Turvey, Stuart E; Durandy, Anne; Fischer, Alain; Fung, Shan-Yu; Geha, Ralf S; Gewies, Andreas; Giese, Thomas; Greil, Johann; Keller, Bärbel; McKinnon, Margaret L; Neven, Bénédicte; Rozmus, Jacob; Ruland, Jürgen; Snow, Andrew L; Stepensky, Polina; Warnatz, Klaus

Titel des Beitrags:
The CARD11-BCL10-MALT1 (CBM) signalosome complex: Stepping into the limelight of human primary immunodeficiency.

Abstract:
Next-generation DNA sequencing has accelerated the genetic characterization of many human primary immunodeficiency diseases (PIDs). These discoveries can be lifesaving for the affected patients and also provide a unique opportunity to study the effect of specific genes on human immune function. In the past 18 months, a number of independent groups have begun to define novel PIDs caused by defects in the caspase recruitment domain family, member 11 (CARD11)-B-cell chronic lymphocytic leukemia/lymphoma 10 (BCL10)-mucosa-associated lymphoid tissue lymphoma translocation gene 1 (MALT1 [CBM]) signalosome complex. The CBM complex forms an essential molecular link between the triggering of cell-surface antigen receptors and nuclear factor ?B activation. Germline mutations affecting the CBM complex are now recognized as the cause of novel combined immunodeficiency phenotypes, which all share abnormal nuclear factor ?B activation and dysregulated B-cell development as defining features. For this "Current perspectives" article, we have engaged experts in both basic biology and clinical immunology to capture the worldwide experience in recognizing and managing patients with PIDs.
caused by CBM complex mutations.

Zeitschriftentitel / Abkürzung:
J Allergy Clin Immunol

Jahr:
2014

Band:
134

Heft / Issue:
2

Seiten:
276-84

Sprache:
eng

Pubmed:

Print-ISSN:
0091-6749

TUM Einrichtung:
Institut für Klinische Chemie und Pathobiochemie

Occurences:
- Einrichtungen > Fakultäten > Fakultät für Medizin > Kliniken und Institute > Institut für Klinische Chemie und Pathobiochemie > 2014

entries: