A human immunodeficiency caused by mutations in the *PIK3R1* gene

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The Journal of clinical investigation. 2014;124(9):3923-8

Heterozygous splice mutation in *PIK3R1* causes human immunodeficiency with lymphoproliferation due to dominant activation of PI3K

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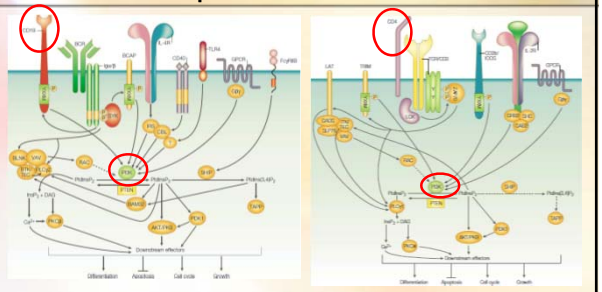
The Journal of experimental medicine. 2014;211(13):2537-47

PhosphoInositide 3-Kinase



- are important regulators of adaptive immunity
- have been linked to an extraordinarily diverse group of cellular functions, including cell growth, proliferation, differentiation, motility, survival and intracellular trafficking

Too little PI3K activity leads to immunodeficiency, whereas too much activity leads to autoimmunity and leukaemia

PIK3R1 - brist

- Immunbrist – ffa återkommande infektioner i sinus/lungor
- Lymfoproliferation
- Ökad mottaglighet för EBV och CMV
- Ökad risk för inflammatoriska sjukdommar
- Ökad risk för lymfom

  - Minskad antal naiva T-celler
  - expansion of CD8 celler
  - Dålig immnuglobulinproduktion – behöver IVIG/SCIG