Term paper

1. What is already known on this topic?

* Subcutaneous panniculitis like T-cell lymphoma (SPTCL) is a unique type of lymphoma, manifests with skin nodules, present in younger age at onset and, unlike other lymphomas, responds well to immunosuppressive therapy.
* Pathogenesis of the disease is unknown. Genetic predisposition is suspicious to be an underlying cause of the disease

1. What this study adds

* Recurrent germline, loss-of-function, missense *HAVCR2* mutations (p.Tyr82Cys and p.Ile97Met) were found in 60% in SPTCL patients.
* Since SPTCL doesn’t have definite diagnosis criteria, sometimes it is difficult to distinguish with lupus panniculitis, identification of germline *HAVCR2* mutation can help making the diagnosis of this lymphoma and repression of TIM-3 (HAVCR2 coding for TIM-3 protein) might be effective for the treatment target.

1. How the data were analyzed

Pick 3 figures and describe the followings

Figure 1a

1. What is question that this figure tried to answer?

* What are the genotype and ethnicity of the affected and families (if available)?
* Among the various ethnicity, who is the ancestor?

1. What data have been generated to answer the question?

* Genotypes and pedigree of the patients and families
* Haplotype of the family members

1. What analysis have been done to get to the conclusion?

* Whole exome sequencing of the samples.
* Haplotype reconstruction from genotyping data using six microsatellite markers to suggest founder effect.

Figure 1b

1. What is question that this figure tried to answer?

* How conserved are the variants we identified in SPTCL patients?

1. What data have been generated to answer the question?

* DNA sequences among species.

1. What analysis have been done to get to the conclusion?

* Comparison of the DNA sequence in the IgV-like domain region among species.

Figure 1c

1. What is question that this figure tried to answer?

* How important are the variants identified in the SPTCL patients?

1. What data have been generated to answer the question?

* Location of the position of the variants in the gene.

1. What analysis have been done to get to the conclusion?

* Study gene structure and locate variants in the domain of the gene.