Stellingen behorende bij het proefschrift

Clinical and Immunological Diversity of Recombination Defects

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1. Coding joint analysis is a powerful tool to identify the type of recombination defect.
   (This thesis)

2. NHEJ defects need also to be considered in patients with reduced circulating numbers of naive T cells and B cells, who presented with milder clinical symptoms than classical T-B-SCID.
   (This thesis)

3. The clinical outcome of an individual RAGD patient, depends on a complex interplay between the (limited) immune receptor repertoire, (auto)antigen exposure, and the specificity of antigen receptors present at the time of immune activation.
   (This thesis)

4. AON treatment is a promising approach to treat patients with intronic splice-site mutations.
   (This thesis)

5. Studying the immune receptor repertoire with next generation sequencing demonstrated that AT, NBS and a subgroup of CVID patients have reduced immune receptor diversity.
   (This thesis)

6. Newborn screening for SCID with both TREC and KREC ensures early detection of B-and T cell deficiencies.
   (Chan *et al. J Allergy Clin Immunol.* 2005;115:391-8 and Nakagawa *et al. J Allergy Clin Immunol.* 2011;128:223-225)

7. As important as techniques to produce the NGS data that address biological questions are, analytical approaches are equally critical for successful interpretation of those data.
(Koboldt *et al. Cell.* 2013;15:27-38)

8. The importance of DNA repair is evident from the large investment that cells make in DNA repair enzymes.
(Alberts *et al.* Molecular Biology of the Cell, 4th edition)

9. Disturbance of translational regulation is one of the explanations why patients with ribosomal deficiencies predominantly have anemia.
(Horos *et al. Blood.* 2012;119:262-272)

10. You are never too old to set another goal or dream a new dream.
(C.S. Lewis, "Chicken soup for the soul", 1993)

11. In the end, V(D)J recombination is just cutting and pasting of DNA.