Stellingen behorende bij het proefschrift

“For gut development: an act of balance”

Next generation sequencing and copy number variation profiling in esophageal atresia

1. The presence of a genetic syndrome is a risk factor for esophageal atresia. This thesis

2. Inherited rare and private mutations and copy number variations can shift the balance from normal to abnormal development. This thesis

3. Genomic sequencing and high resolution copy number variation profiling will identify previously known and new genetic syndromes in many previously “VACTERL association” diagnosed patients. This thesis

4. Comparison of monozygous twin DNA is an excellent method to optimize next generation sequencing. This thesis

5. In view of the genetically heterogeneous etiology of EA/TEF, only worldwide collaboration will permit us to identify patients who share the same causal factor. This thesis

6. Genomic data matures like fine wine, it is best to wait a few years before reanalysis.

7. We can only speculate on our future knowledge on what is or isn’t “pathogenic” variation. Therefore, true “informed consent” of parents is impossible.

8. At first sight, only failure to close the bow doors triggered the capsizing of the “Herald of free Enterprise”. However, closer examination revealed that many cumulative acts resulted in this catastrophe.

9. If you torture the data long enough, it will confess. (Ronald Coase, economics Nobel Prize laureate 1991)

10. The larger part of research budgets is spent on investigating what causes a disease, more budget should be allocated to disease prevention and dissemination of what is already known.

11. A PhD trajectory is like a long distance race, you start optimistically, halfway you wonder if you are going to make it to the finish and in the end you get a sense of accomplishment.