Stellingen behorende bij het proefschrift

Congenital Diseases of the Intestine

1. Efforts to advance therapy for patients with CSBS, MMIHS or HMIA are hampered by our limited understanding of their pathogenesis. *(this thesis)*

2. Finding disease-causing genes has been greatly facilitated by the rapid progress in technologies such as next generation sequencing (NGS), and further testing of identified candidate genes by genome editing techniques (in animal models) is currently making forward genetic studies possible. *(this thesis)*

3. ACTG2 is the main actin isoform expressed in intestinal smooth muscle cells, and changes affecting its structure lead to the dominant form of MMIHS. *(this thesis)*

4. Loss-of-function mutations in *LMOD1* and *MYLK* cause recessive MMIHS, confirming MMIHS is a heterogeneous disease of the visceral organs, with multiple patterns of inheritance. *(this thesis)*

5. Genetic findings in CSBS patients, including the identification of *CLMP* and *FLNA* disease-causing variants, represent a first step to identify major processes required for intestinal development and elongation. *(this thesis)*

6. Zebrafish is an excellent animal model to study the intestinal lumen formation, and has the potential to shed light on the disease mechanism of HMIA. *(this thesis)*

7. The field of NGS development and applications is a fast-moving area of research, which makes this an exciting time for genomic studies. (*Metzker, Nature Reviews Genetics, 2010*)

8. With further technological improvements and increasing success rates, prenatal and preimplantation diagnosis of genetic disorders will become commonplace. (*Vermeesch et al., Nature Reviews Genetics, 2016*)

9. The field of tissue engineering is ripe for expansion and it requires training of a generation of materials scientists and chemical engineers. (*Vacanti, Vacanti, Principles of Tissue Engineering, 2007*)

10. I like the freedom of research. Plus, if I fail in science, I know I can always survive because I have an M.D. This has been my insurance policy. (*Shinya Yamanaka*)

11. Anyone who has never made a mistake has never tried anything new. (*Albert Einstein*)

Danny Halim
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