Paediatric cardiomyopathies

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Stellingen behorend bij het proefschrift

Paediatric cardiomyopathies

An evolving landscape of genetic aetiology and diagnostic applications

1. Compound heterozygous or homozygous *MYBPC3* variants should be considered in a neonate presenting with severe hypertrophic or noncompaction cardiomyopathy, even in the absence of a positive family history for cardiomyopathy or sudden cardiac death. (this thesis)
2. In children with cardiomyopathy, exome sequencing is preferable to targeted next-generation sequencing. (this thesis)
3. In children with cardiomyopathy and consanguineous parents, combining haplotype sharing analysis with exome sequencing provides a powerful strategy to identify novel genes. (this thesis)
4. Genetic testing should be recommended for children with chemotherapy-induced dilated cardiomyopathy or (suspected) myocarditis. (this thesis)
5. Novel gene prioritization tools such as GeneNetwork Assisted Diagnostic Optimization (GADO) are urgently needed in order to increase the diagnostic yield of whole exome/genome sequencing. (this thesis)
6. Patients carrying a heterozygous loss-of-function variant in *ALPK3* are at risk of developing hypertrophic cardiomyopathy and should be offered periodic cardiac screening. (this thesis)
7. Transition from dilated to hypertrophic cardiomyopathy appears to be unique to biallelic *ALPK3*-related cardiomyopathy. (this thesis)
8. The identification of *superoxide dismutase 2* (*SOD2*) as a novel gene involved in human neonatal cardiomyopathy illustrates that pathophysiological mechanisms seen in common cardiovascular diseases are also involved in rare disorders. (this thesis)
9. Een arts die ook wetenschappelijk onderzoek doet, is een onmisbare schakel in translationeel onderzoek.
10. "Een groot mens is hij, die zijn kinderhart nooit verliest" – Meng-Tse, Chinees filosoof
11. "Ook vroeger was niet alles zoals het vroeger was" – Herman van Veen, Nederlands muzikant, schrijver en schilder

Johanna C. Herkert, 16 oktober 2019