

Hirschsprung Disease:

Development & Treatment Avenues

1. Somatic mutations are unlikely to be causative for Hirschsprung disease, due to the developmental pattern of the enteric neural crest (this thesis)
2. A threshold decrease in *KIF1BP* expression is likely necessary to develop Goldberg-Shprintzen syndrome (this thesis)
3. Large Copy Number Variations occur more in Hirschsprung patients with additional developmental anomalies and an unknown genetic aetiology (this thesis)
4. Induced pluripotent stem cells from patients are likely to be the most viable cell source for cell transplantation therapy. Safety and genomic stability of these cells, however, remains questionable (this thesis)
5. In order to restore anal sphincter function to a wider range of patients with fewer negative side-effects, the development of a technology-based treatment may be superior to cell therapies in the near future (this thesis)
6. The effect of any single factor contributing to a complex disease may be obscured or confounded by other contributing factors.
7. The ultimate goal of therapy is a subjective improvement of symptoms that increases the individual's quality of life.
8. By bringing together experts from different disciplines we can find the solutions for today's challenges.
9. Machine learning is swiftly infiltrating many areas within the healthcare industry, with significant potential to transform the medical landscape.
10. Science is not a business. Knowledge is not a commodity to be bought and sold, it does not lose value the more people that obtain it.
11. If at first you don't succeed try, try and try again.
(Robert the Bruce of Scotland)