

# The Genotype-Phenotype Correlation in Pompe Disease Studied from an Enzymatic and Molecular Perspective

by

Monica Y. Niño M.

1. Cultured skin fibroblasts are the best sample source for the enzymatic diagnosis of Pompe disease because of the low false positive and false negative outcome rates, and for discriminating between classic infantile and less progressive phenotypes (*this thesis*).
2. The inclusion of a clinical severity rating in the Pompe disease *GAA* variant database ([www.pompevariantdatabase.nl](http://www.pompevariantdatabase.nl)) provides an invaluable tool for diagnosis, prognosis, and treatment of Pompe disease including the future development of personalized medicine (*this thesis*).
3. The influence of putative modifying factors is particularly large for the c.-32-13T>G variant as compared to other *GAA* variants (*this thesis*).
4. The *ACE* I/D polymorphism alone does not explain the large variation in disease severity and response to ERT observed among patients with Pompe disease and the same c.-32-13T>G / null *GAA* genotype (*this thesis*).
5. The identification of factors that modify the clinical course of Pompe disease will lead to a better understanding of the disease process, and may provide novel disease markers and therapeutic targets (*this thesis*).
6. Governments, United Nations agencies, civil society, research organizations and the food industry should work together to develop and promote strategies for sustainable food production (Alsaffar AA. *Food Sci Technol Int*, 2016).
7. "Advances in studying the genomic traits driving differential pharmacogenomics will undoubtedly enhance individualized treatments for a wide variety of diseases" (Moscoso CG et al. *Ann N Y Acad Sci*, 2020).

8. With advancing screening technology, a future choice will be created between carrier detection or newborn screening (Yao R., Goetzinger K.R. *Clin Lab Med*, 2016 & Gelb M.H. et al. *Int J Neonatal Screen*, 2019).
9. Future advances in prenatal gene therapy have the potential to overcome some of the current limitations of efficient postnatal gene therapy (Peranteau WH, Flake AW. *Mol Diagn Ther*, 2020).
10. "Cultured meat could be an alternative for consumers who want to be more responsible but do not wish to change their diet". However, current research is still required to optimize cell culture methodology, impact on health, and safety (Chriki S, Hocquette JF. *Front Nutr*. 2020).
11. Covid-19 is the first major pandemic of our generation, and we must seek the hidden opportunities to reflect and react as a global society, challenging our position in the world as an opportunity for social transformation (Ahmad A. et al. *BMJ*, 2020).