Investigating the genetic complexity of glaucoma

Lo Faro, Valeria

DOI:
10.33612/diss.216689872

Propositions accompanying the dissertation entitled:

Investigating the Genetic Complexity of Glaucoma

1. Standard genome-wide association studies focus on common variants and do not capture the entire range of genetic variations such as structural and copy number variations. (This thesis)

2. Genetic risk approaches do not fully account for private variants, structural variants, and variants whose effect is modified by environmental factors. (This thesis)

3. The under-representation of non-Caucasian populations in genetic studies is problematic for scientific and ethical reasons. The effects of gene variants that are present only in the underrepresented ethnic groups remain unknown. (This thesis)

4. There is an urgent demand to produce data and polygenic scores in non-Caucasian populations in order to explore and characterize the extent to which transethnic transferability of polygenic scores can be implemented. As such, African genomes are finally getting the attention they deserve. (This thesis)

5. "From an early age, I came to think of myself as others thought of me: chronically ill. Every skinned knee and runny nose were treated as if it were life-threatening." (GATTACA)

6. It should be clear that genetics represents only one of the contributors to individual disease risk. Therefore, for multifactorial traits, the genetically defined risk must not be considered deterministic.

7. New counselling methods in the communication of genetic risk are needed to encourage individuals to modify and undertake long-term behavioural changes required for healthy ageing.

8. Understanding how genetic risk and social inequality interact in influencing health disparities will be critical to improve public health.

9. “A journey of a thousand miles begins with a single step.” (Laozi)

Valeria Lo Faro