Knowledge multiplies when shared — when calling things by their right name: improving the validation and exchange of genetic data in research and diagnostics

Fokkema, I.F.A.C.

Stellingen behorende bij het proefschrift getiteld

Knowledge multiplies when shared — when calling things by their right name

Improving the validation and exchange of genetic data in research and diagnostics

1. The HGVS Nomenclature was designed with the human user in mind. As such, it has become a very human-like standard: filled with inconsistencies and ambiguities, and difficult to fully comprehend. (this thesis)

2. The "variant type juggling" shown in Mutalyzer 3 reveals missing prioritization rules in the HGVS Nomenclature. This raises the question whether human-readable prioritization rules can ever be exhaustive. (this thesis)

3. By ignoring existing standards and left-aligning variant positions, the Variant Call Format (VCF) caused problems in genetic diagnostics worldwide, as tools that convert between VCF and HGVS do not always correct for this. (this thesis)

4. If those generating genetic data, submitted to databases as much as they queried them, the money spent on digging up data buried in the literature could have been spent on finding treatments, instead. (this thesis)

5. Authors and publishers owe patients and their families the relatively small effort of at least applying the variant nomenclature standards to their data. (this thesis)

6. When accepting projects using unfunded DNA variant databases, funding providers should set aside part of the budget to compensate these databases, supporting open science by keeping them open for all. (this thesis)

7. Data models should represent the real world as closely as possible; every simplification reduces their usefulness. (this thesis)

8. When something is hard to do right but looks simple, it is usually done poorly — making it look difficult may well be the main benefit of the Variation Representation Specification (VRS).

9. While much of science depends on bioinformatics, some wet-lab scientists claim that bioinformatics is not a science in itself. Yet, writing code is much like running experiments in parallel all day in a lab built and equipped entirely by the developer themselves.

10. It should be possible for someone to be both first and last author on a single paper.

11. Studies show that our quality of life depends more on our mental health than our physical health. Yet, our healthcare system — as well as society at large — remains largely focused on physical wellbeing.

12. If you think you're always right, you'll never learn anything — and as such, you'll soon always be wrong.