Proc. Assoc. Advmt. Anim. Breed. Genet. 22:253

PRECISION MEDICINE: REALISTIC EXPECTATIONS FOR PREDICTION OF RISK TO HUMAN COMPLEX DISEASE

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SUMMARY

I will review the progress and prospects of risk prediction for disease in people. Fundamental to this progress is that data sharing is now common-place for both association study summary statistics and of individual-level measures.

GENETIC RISK PREDICTION

The methodology of genetic risk prediction of human disease parallels genetic evaluation in livestock. However, fundamental differences reflect the data structure available for generating and validating predictors, and that the prediction goal is of an uncommon binary phenotype of an individual, rather the mean value of a quantitative trait in the next generation. Genetic predictors of common complex genetic diseases, can never be diagnostically accurate for an individual, but genetic risk stratification could have clinical utility. For example, risk stratification could identify a high-risk class that includes the majority of those who will become affected in their lifetime (high sensitivity) even though the majority of those in this high-risk class will not be affected (poor specificity). Accurate prediction of a phenotype, requires the genetic predictor to be enhanced to include non-genetic risk factors. The genomics era allows the inclusion genomic biomarkers, which could reflect the downstream consequences disease and of non-measured environmental risk factors.

I will review the progress and prospects of risk prediction for disease in people. Fundamental to this progress is that data sharing is now common-place for both association study summary statistics and of individual-level measures. For example, the UK Biobank (Sudlow et al. 2015; www.ukbiobank.ac.uk) study of 500,000 people with deep phenotyping and genome-wide genotype data now presents opportunities for quantitative genetics methods common in livestock to be applied to human data, and provides new opportunities for cross-fertilisation of ideas between disciplines. While disease risk prediction receives much hype in the era of personal or precision medicine it is important to not to oversell what can be realistically achieved.

REFERENCES

Sudlow C, Gallacher J, Allen N, Beral V, Burton P, Danesh J, et al. UK Biobank: An Open Access Resource for Identifying the Causes of a Wide Range of Complex Diseases of Middle and Old Age. PLoS Med. Public Library of Science; 2015;12: e1001779.