

Genetic research in a biological sample: pharmacogenomics for vulnerable populations.

S. Gaudi (1), A.L. Knellwolf (1), F. Macciardi (2, 3), S Vella (1) simona.gaudi@iss.it

(1) National Institute of Health, Rome. Italy (2) Università degli Studi di Milano, Milano. Italy (3) University of California Irvine, UCI. USA

Acknowledgement:

NEAT- European AIDS treatment network of excellence (LSHP-CT-2006-037570-FP: 6)

Introduction The investment in, and adoption of, innovative science and technology is crucial to improving the health of population and stimulating emerging economies and developing countries. These efforts are crucial for breaking the cycle of dependence on industrialized countries. Emerging economies have chosen to publicly fund large-scale national human genotyping initiatives to explore human genomic variation in their respective populations. Following the completion of Human Genome Sequencing a large-scale genotyping projects have been initiated in developed countries to explore the relationship between SNPs and population variance in disease predisposition (early diagnosis) and drug response (pharmacogenomics). Genomic medicine and pharmacogenomics have the potential to provide cost saving in drug development, to reduce health-care cost, to stimulate growth and investment within the local public and private sector, and to uncover genetic diversity that is relevant to drug response or disease predisposition.

Methodology One of the major development in the field of the Human Genetics has been the arrival of the Genome-Wide Association Studies or GWAS. Similar to positional cloning, GWAS do not need any a priori hypothesis of the underlying pathology. However, we are also at the entry of a new era: for the first time in the history of biomedicine, GWAS provide us with a powerful and accurate tool to tackle the complete genome for the disease gene search entirely without segregation information. This innovative technology has the potential to generate a key resource for local researchers to understand disease susceptibility and variation in drug responses, which will contribute to the goal of developing public health genomics amongst emerging economies. Drugs that have been developed and tested in Caucasian population of industrialized countries would have the same therapeutic index (benefit/risk ratio) in vulnerable populations?

Results Emerging genomic technologies, such as sequencing and genotyping, can potentially be harnessed to improve health and reduce the burden of disease. Mexico, India, Thailand and South Africa have publicly founded large-scale national human genotyping initiatives to explore human genomic variation in their respective populations. Emerging knowledge about local human genomic variation in these countries can also be harnessed by the local domestic public and private sector towards creating cost efficiencies and improving their drug development pipelines.

Conclusion

New generation of genomic scientists who have additional parameters in mind, such as building infrastructure and training human resources is essential for effective translation of genome-based science and technologies in vulnerable populations. One of the major challenges in the application of genomic medicine in vulnerable population, involves the limited regulatory infrastructure. Genomic medicine is redefining how both developed and developing countries need to work together in the application of new knowledge to improve global health.

Key words: genomic medicine, GWAS, vulnerable populations, global health