

Stellingen behorend bij het proefschrift

Genetic Determinants of Non-syndromic Hearing Impairment

1. Genetic hearing impairment is extremely heterogeneous, and the number of genes involved may total up to 300 or 1% of all human genes. – *After: Nance WE. Ment Retard Dev Disabil Res Rev 2003; 9: 109-119.*
2. The Pakistani population is a valuable resource for mapping non-syndromic hearing impairment genes, with ~29 loci identified in Pakistani families, including five loci that are described in this thesis.
3. The gene-specific prevalence rates among Pakistani families for functional variants in the genes *GJB2*, *TMC1* and *TMIE* are low, and must be explained by random genetic drift (this thesis).
4. Bioinformatics tools, such as multiple sequence alignment and transmembrane domain prediction, should be used in order to predict if a variant is functional (this thesis).
5. For new gene discovery in hearing impairment to be clinically useful, studies on gene-specific prevalence rates, spectra of sequence variants, phenotypic description and functional analyses are required (this thesis).
6. Phenotypic description for non-syndromic hearing impairment loci aids not only gene mapping but also clinical management (this thesis).
7. DNA sequence analysis to discover population polymorphisms that may predispose to regionally specific diseases is one of the most promising biotechnologies for improving health in developing countries. – *Daar AS et al. Nat Genet 2002; 32: 229-232*
8. The study of head and neck tumors through microarray data has great potential in the elucidation of progression of molecular disease, the prediction of the occurrence of tumors and morbid outcomes, and the identification of altered biologic pathways that are novel therapeutic targets. – *Choi P and Chen C. Cancer 2005; 104: 1113-1128.*
9. The majority of studies find that racial and ethnic disparities [in health care] remain even after adjustment for socioeconomic differences and other healthcare access-related factors. – *Institute of Medicine. "Unequal treatment: confronting racial and ethnic disparities in health care" Washington DC: National Academies Press, 2003.*
10. The clinical academicians, the physicians who care for patients and also spend time directing their own laboratory effort, represent the critical elements to facilitate clinical-basic collaborations and promote translational studies. – *After: Holcombe RF. Acad Med 2005; 80: 905-907.*
11. Philippine mango is the best mango in the world.

REGIE LYN PASTOR SANTOS, 2006

Genetic Determinants of Non-syndromic Hearing Impairment

REGIE LYN PASTOR SANTOS