



# Definitions, Protocols and Guidelines in Genetic Hearing Impairments

*A. Martini, Alessandro Martini, Andrew Read, Dai Stephens, Manuela Mazzoli*

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This book brings together many of the main conclusions of the European Concerted Action Programme on Genetic Hearing Impairment (HEAR). It is split into four sections, covering definitions, protocols, genotype/phenotype relationships and important websites. The section on definitions enables all those approaching the problems of Genetic Hearing Impairment from different backgrounds to communicate in the same language and understand what each is doing more clearly. The definitions are of Audiological, Vestibulological, Epidemiological and Genetic terms, together with specific terms associated with particular craniofacial abnormalities.

The second section comprises protocols for the minimal set investigation of patients and their family members with genetic hearing impairment. Relatively little work has been done in the past on the balance of disorders which may be associated with Genetic Hearing Impairment and a protocol aiming to elucidate some of these factors in a relevant way has been defined. The final chapter in this section deals with how audiologists should relate to genetic laboratories in an attempt to reduce the confusion which has risen in this field in the past.

The third section is concerned with the relationship between genotypes and phenotypes in non-syndromal hearing impairment in the conditions in which the genes have so far been localised and in many cases identified.

The final section deals with the important websites within this field. The most important and the most visited is the Hereditary Hearing Loss website based on Antwerp and managed by Guy Van Camp and Richard Smith.

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