BioISI - Biosystems & Integrative Sciences Institute

BioISI Research Seminar

Contribution of Genetics to Hearing Impairment Diagnosis

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Host: Astrid Vicente (BTR-BioISI)

When: Thursday February 28 - 12h00
Where: Building C2, Room 2.4.16

Hearing loss (HL) is the most common congenital sensory impairment, affecting one in 500 newborns and one in 300 children by the age of 4. HL limits the ability to develop effective auditory and speech capacities as well as the social integration of individuals as valid citizens. The etiology of HL can be classified according to its nature being sensorineural hearing loss (SNHL) our study focus. Congenital HL could be non genetic (acquired) or genetic, syndromic or non-syndromic, being our efforts dedicated to the identification of causative genes and mutations associated to HL. HL can be classified according to the severity being most of our cases severe (71-90 dB HL) and profound (more than 90 dB HL). Management of hearing loss is mainly influenced by the nature, the bilaterality, the severity and the age at diagnosis. Severe to profound bilateral SNHL can be managed by cochlear implantation (unilateral or bilateral), if picked up at early age. High number of genes associated to some HL forms are described, some associated both to syndromic and non-syndromic cases. The most frequent genetic cause of congenital sensorineural hearing loss (SNHL) is disruption of GJB2, the gene encoding Connexin 26 (Cx26), the major component of gap junctions in the cochlea. We present our results for different studies related to genetic etiology of different cases, discussing also its relevance for diagnosis.

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