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SEMINAR PROGRAMME 2018

Monday, 8 January 2017 | 3:00 pm | ICGEB Seminar Room, W building | Padriciano, 99, Trieste, ITALY

Fabio MAMMANO

Director, CNR Institute of Cell Biology and Neurobiology , International Campus "A. Buzzati-Traverso", Monterotondo (RM), ITALY

"Hearing impairment and skin diseases associated with connexin 26 mutations: etiopathogenesis and translational opportunities"

Host: M. Giacca

The *GJB2* gene has an estimated mutation prevalence of 3% in the general population. The encoded membrane protein, connexin 26 (Cx26), is expressed in the inner ear and the skin, together with the closely related connexin 30 (Cx30). Most *GJB2* mutations cause nonsyndromic forms of hearing impairment, which are prevalently autosomal recessive or, more rarely, autosomal dominant, and together affect ~1 in 2000 newborn children. In addition, a certain number of dominant *GJB2* mutations cause syndromic forms associated with an array of rare skin diseases (Bart-Pumphrey syndrome; Hystrix-like ichthyosis with deafness; Keratitis-ichthyosis-deafness syndrome; Keratoderma, palmoplantar, with deafness; Vohwinkel syndrome). Non-syndromic hearing impairment is mainly associated with *GJB2* mutations that cause complete loss of protein function, such as the highly prevalent 35delG, whereas most syndromic forms are causally linked to hyperactive mutant channels. Both classes of mutations represent highly challenging and virtually uncharted translational opportunities. I will discuss the etiopathogenesis of hearing loss linked to *GJB2* mutations and highlight our attempts to treat relevant mouse models using recombinant adeno associated viral vectors with high tropism for inner ear non-sensory cells.

I will also present a promising approach to treat skin disorders by contrasting channel hyperactivity with fully human monoclonal recombinant antibodies selected from a phage library.

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More information at:

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