

# 2025 臺中榮民總醫院院慶國際醫學研討會

## TCVGH International Medical Conference

### AI in Medicine

Future of Healthcare by AI



請於 8 月 15 日前回傳表單

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| Professional Career  | 臺中榮總耳鼻喉頭頸部主治醫師<br>國立陽明交通大學兼任助理教授                           |                         |
| Speech Title   | Clinical Phenotypes of Common Hearing Loss Genes in Taiwan |                         |
| Abstract( 200 words) :<br>In Taiwan, genetic factors cause over two-thirds of newborn hearing loss cases. Understanding the common genes is crucial for timely management. The <b>GJB2 gene</b> is most prevalent, inherited recessively. Its <b>c.109 G&gt;A variant</b> can cause mild-to-moderate loss, sometimes missed by screenings. The <b>c.235delC variant</b> often leads to moderate-to-profound loss, requiring early intervention like hearing aids or cochlear implants. The <b>SLC26A4 gene</b> (also recessive) can cause <b>Pendred syndrome</b> (with thyroid issues) or hearing loss with <b>Enlarged Vestibular Aqueduct (EVA)</b> , making hearing unstable. <b>OTOF gene</b> variations lead to <b>Auditory Neuropathy Spectrum Disorder (ANSO)</b> , where speech discrimination is poor despite varying hearing thresholds. <b>MYO15A gene</b> typically causes <b>severe, non-progressive sensorineural hearing loss</b> from birth. The maternally inherited <b>MTRNR1 gene</b> is critical; its <b>m.1555A&gt;G variant</b> can cause rapid hearing deterioration with <b>aminoglycoside antibiotics</b> , which must be avoided. <b>KCNQ4 c.546C&gt;G variant</b> is associated with early-onset high-frequency hearing loss, tinnitus, and cardiovascular comorbidities in Taiwanese adults.<br>Early genetic testing is vital for accurate diagnosis and personalized treatment plans. |  |                         |