OC1

CLINICAL AND MOLECULAR FEATURES OF TWENTY CHILDREN WITH HYPER-IGE SYNDROME CAUSED BY STAT3 GENE MUTATION IN MAINLAND CHINA

Jiang Liping*, Xiang Qingqing. *Children's Hospital of Chongqing Medical University, Chongqing, China*

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Background and aims STAT3 gene mutation causes autosomal dominant hyper- IgE syndrome (AD-HIES) is a rare primary immunodeficiency disease. We described the clinical and molecular features of 20 AD-HIES patients in Mainland China.

Methods We diagnosed patients with AD-HIES on the basis of NIH score of ≥ 40 points, described the clinical features, sequenced the STAT3 gene of 20 patients, and quantified Th17 cells in peripheral blood of 19 patients by flow cytometry.

Results Among the 20 patients (14 males and 6 females), the onset age ranged from 1 days to 6 months, and the diagnostic age ranged from 1 to 12 years. All patients (100%) had characteristic manifestation of HIES with the range of NIH scores 41–77 points, including recurrent eczema and pneumonia, particularly high serum IgE levels and eosinophilia. Characteristic facial features (100%), bone abnormalities, recurrent skin abscesses (85%), lung abscesses (60%), recurrent oral thrush (55%) were also present in the studied patients. STAT3 hot mutations V637M or R382W/Q were identified in 12 patients (60%), two novel heterozygous missense mutations (T620S and R609G) were identified in 2 patients respectively, and other mutations (C712G, S636F, Q469H, Y705H, V432M, c.1139+5G>T) were identified in other 6 patients. The percentage of Th17 cells in CD4+ T cells (CD3+CD8IL-17A+/CD3+CD8 cells) of 19 patients with STAT3 mutation was significantly decreased or deficient as compared with healthy controls.

Conclusions Children with repeated *Staphylococcus aureus* pneumonia, cold skin abscess, fungal infection, eczema, increased serum IgE level and eosinophil, should be alert to AD-HIES. Detection of Th17 cell by flow cytometry assist clinical diagnosis when the NIH score ≥ 40 points. Further STAT3 gene analysis can confirmed the diagnosis. Early diagnosis and active prevention of infection can significantly improve the quality of life of patients.

OC2

A NEW PEDIATRIC EDUCATIONAL MODEL: NEONATAL APPLICATION OF WORD CLOUD ANALYSIS AND SINGLE WORD SUMMARISATION

Roy K Philip*. *Graduate Entry Medical School (GEMS), University of Limerick, Limerick, Ireland. University Maternity Hospital Limerick (UMHL), Limerick, Ireland*

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Background Word cloud (WC) is the visual representation of a collection of text documents that uses various font sizes, colours, and spaces to arrange and depict significant words. WC generated from spontaneously articulated views by medical students as a single word summarisation (SWS) could assist in medical teaching. SWS could also assist pediatric teaching faculty to foster better student engagement. There is a paucity of published material in pediatric medical education using the WC analysis technologies.

Methods A descriptive study was conducted in the neonatal intensive care unit (NICU) of University Maternity Hospital

Limerick (UMHL) for five years from October 2012 to September 2017. One faculty member prospectively recorded the SWS of neonatology by medical students. Hospital audit committee approved study as a quality improvement project (QIP) in clinical teaching. Online WC generator under an open source license was used to compute the WC and an automated layout algorithm for positioning words without overlap was chosen.

Results Total of 268 SWS were generated from 268 graduate entry medical students who had neonatal rotations over five years. Words reflecting 'abstract' concepts predominated (example -rewarding) (182/268) compared to 'concrete' concepts (example- incubator) (86/268) with $p < 0.005$. Numerical predominance with maximum repeatability was noted for the following fifteen words: intensive, ventilation, nutrition, emotional, high-risk, sepsis, premature, outcomes, risk, mortality, rewarding, nursing, feeding, sensitive and careful. Feedback data showed SWS and WC generation as unique 152/268 (56.7%), stimulating 142/268 (52.9%) and creative 130/268 (48.5%). Interestingly 72/268 (26.8%) found it as a fun. A relatively small proportion found it not interesting 20/268 (7.4%), boring 8/268 (3%) and waste 4/268 (1.5%). Power point slides of WC aided students to reflect on the visual impact of their chosen words.

Conclusion Visualization of medical student-generated SWS could stimulate reflection and clinical discussion. SWS could assist teaching faculty to foster better student engagement. This narrative teaching methodology with computer aided WC enhancement has the potential transferability to other clinical disciplines.

OC3

A HOST-RESPONSE ASSAY DISTINGUISHES BETWEEN SIMPLE INFLUENZA PATIENTS AND INFLUENZA PATIENTS WITH BACTERIAL COINFECTION

^{1,2}Adi Klein*, ³Meital Paz, ³Kfir Oved, ³Asi Cohen, ^{2,3,4}Ellen Bamberger, ^{2,4}Isaac Srugo, ^{2,4}Israel Potasman, ³Eran Eden, ³Liran Shani. ¹Hillel Yaffe Medical Center, Hadera, Israel; ²Technion-Israel Institute of Technology, Haifa, Israel; ³Me Med, Tirat Carmel, Israel; ⁴Bnai-Zion Medical Center, Haifa, Israel

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Background Identifying bacterial coinfection in influenza patients can be difficult as the symptoms of simple influenza versus mixed infections are often similar, leading to antibiotic overuse. A new host-response assay (ImmunoXpert™) that integrates the levels of three proteins (TRAIL, IP-10, and CRP) was shown to exhibit high performance in distinguishing between bacterial and viral disease in two double-blind validation studies. Here we sought to evaluate its ability to differentiate between simple influenza and influenza with bacterial coinfection.

Material/Methods The study population included 653 febrile pediatric and adult patients prospectively recruited in the 'Curiosity' study. Patient etiology (simple viral versus mixed infection) was determined by unanimous expert adjudication based on comprehensive clinical, laboratory and radiological assessment. Influenza strains (A or B) were detected using multiplex PCR applied to nasal swabs (Seeplex-RV15). We compared the expert panel diagnosis with the assay that gives three possible outcomes: viral, bacterial (including viral with bacterial coinfection) or equivocal. An equivocal outcome does not provide diagnostic information and is observed in ~10% of cases.