Joint Event

36th World Pediatrics Conference

37th International Conference on

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August 07-08, 2023

Webinar



Scientific Tracks & Abstracts

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Novel causative mutation in the gene for Galloway-Mowat syndrome has been identified. Osgep (c.2S G>A p.GLySer) has not been reported in the international database - Report of case and literature review

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Introduction: Galloway- Mowat syndrome is a rare hereditary renal, neurological disease characterized by microcephaly, intellectual disability, hiatus hernia, skeletal anomalies, and nephrotic syndrome. It appears to be transmitted as an autosomal recessive trait. Recently, novel causative mutations for this disease have been identified in the gene-encoding subunit OSGEP. The gene variant has not been reported before in the international database.

Case Presentation: A twenty months old Egyptian with working diagnosis of Galloway- Mowat syndrome caused by OSGEP gene (c.25 G>A p.GlySer). She was born at term by caesarean section due to twin delivery. Birth weight was 2700g. She was born with normal head circumference and weight. At the age of 3 months, mother noticed that her head circumference is not increasing compared to her twin, her current HC <3rd centile. This girl displayed various features of facial dysmorphism (microcephaly, deeply sited eyes, and high arched palate). In addition, she has spasticity, hyperreflexia, truncal hypotonia, Global developmental delay, failure to thrive and epileptic disorders. Renal ultrasound revealed bilateral early to grade 1 renal parenchymatous pathological changes. Her serum creatinine levels were 17 umol/L (low). The segregation analysis showed that both parents and her twin are carriers which supports that the variant of OSGEP is likely to be pathogenic.

Methods: This study was designed as a case report using patient clinical manifestation with a literature review, together with family study through segregation analysis that can yield robust data to re-classify a variant of unknown clinical significance.

Results: The OSGEP gene (c.25 G>A p.GlySer) is most likely pathogenic from the patient phenotype and family segregation data. However, gene functioning is the gold standard method to classify this variant which is still under process.

Conclusions: We report a familial Galloway-Mowat syndrome caused by the OSGEP gene (c.25 G>A p.GlySer) with both parents and her twin carrying a novel heterozygous. She displayed various features; microcephaly, deeply sited eyes, high arched palate, spasticity, hyperreflexia, truncal hypotonia, Global developmental delay, failure to thrive and epileptic disorders.

Biography

Aisha Khameis Ahmed is a third-year pediatric resident in the department of pediatrics at Fujairah Hospital, UAE. She completed MBBS from the UAE University. Before joining the pediatric residency program, she worked as a General practitioner for over seven years in pediatrics.

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The immunoregulation effect of TFEB in dendritic cells on asthma

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Introduction: Asthma is a common chronic disease in children and Dendritic Cells (DCs) play a crucial role in the immunoregulation of asthma. It's reported that overexpression of the Transcription Factor EB (TFEB) alters lysosomal activity and function, enhances MHC II antigen presentation, activates CD4+ effector T cells (Teffs), thus promotes immune activation. Therefore, TFEB may play a critical role in DCs antigen presentation and Teffs activation. However, the immunoregulation role of TFEB in asthma has not been reported.

Methods: Peripheral blood was collected from asthmatic children and TFEB mRNA was detected. A House Dust Mite (HDM)-induced asthma model was established to detect the TFEB, the MHC II and costimulatory molecules (CD40, CD80 and CD86) in DCs and the CD4+ effector T cells (Teffs), including Th1, Th2 and Th17 *in vivo*. After TFEB was inhibited, the expression levels of MHC II and costimulatory molecules (CD40, CD80 and CD86) in DCs, the Teffs and asthma phenotype were detected. Further, the specific mechanisms were further explored.

Results: TFEB mRNA expression levels in peripheral blood of asthmatic children were significantly higher compared with healthy controls. *In vivo* and *in vitro* experiments showed that TFEB expression levels in lung tissues and DCs of asthmatic mice increased significantly after HDM treatment. Inhibiting TFEB expression resulted in a decrease in MHC II and CD40 expression in DCs, as well as a decrease in Th1, Th2 and Th17 cell subsets. Meanwhile, inhibiting TFEB expression levels decreased airway hyper responsiveness, airway inflammation, serum IgE, eosinophil and total cell count in alveolar lavage fluid in the asthma model [Figure 1].

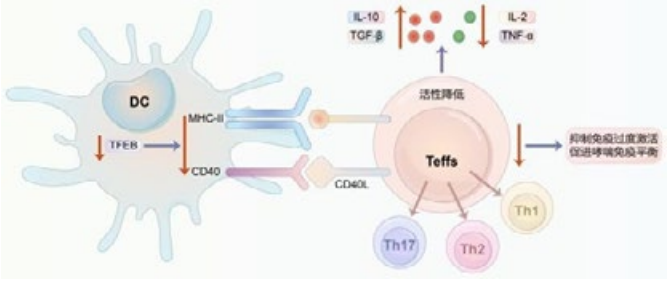

Figure 1. The immune balance in asthma promoted by down-regulation of TFEB in Dendritic Cells (DCs)

Conclusions: TFEB expression is increased in asthma. Inhibiting TFEB expression levels in asthma can protect against immune over activation by suppressing MHC II and CD40 in DCs and reducing the activation of Teffs, thus playing a protective role in asthma.

Biography

Fengxia Ding is a respiratory medicine physician and pediatrician from Children's Hospital of Chongqing Medical University, one of the top three children's hospitals in China. Now she is working as a post doctor in Great Ormond Street Institute of Child Health, University College London (UCL). She has been working as a pediatrician for 7 years in Children's Hospital of Chongqing Medical University and there are more than 19,000 outpatient pediatric patients visit she every year. She got a patent in respiratory diseases and she has over 30 publications focusing on respiratory disease and asthma. She has presented her clinical and research work at many conferences and got many grand's on her researchers.

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Regulation of metabolic reprogramming in alveolar macrophages alleviates lipopolysaccgaride-induced acute respiratory distress syndrome

Bo Liu*, **Fengxia Ding**, **Yan Li**, **Hongbo Li**, **Yonggang Li**, **Chun Wu** and **Zhengxia Pan**
Chongqing Medical University, China

Introduction: Acute Respiratory Distress Syndrome (ARDS) is a common clinical critical illness with a high mortality rate and currently lacks effective prevention and treatment measures. Imbalance in alveolar macrophage polarization plays a crucial role in the occurrence and development of ARDS, but the specific mechanisms underlying alveolar macrophage polarization are still unclear.

Methods: *In vivo* experiments were conducted using intratracheal administration of LPS to establish an ARDS mouse model, while *in vitro* experiments utilized LPS-induced MH-S mouse alveolar macrophages to observe changes in metabolism and phenotype during ARDS. The therapeutic effects of the addition of the glycolysis inhibitor 2-DG were observed and the specific mechanisms were explored.

Results: In LPS-induced ARDS mice, significant inflammatory responses and lung tissue damage were observed, accompanied by an increase in glycolysis levels. The addition of 2-DG markedly alleviated LPS-induced lung injury and reduced inflammation. Mechanistically, LPS induction increased glycolysis levels in alveolar macrophages, promoting polarization towards the M1 pro-inflammatory phenotype. Inhibiting glycolysis shifted alveolar macrophages from the M1 pro-inflammatory phenotype to the M2 anti-inflammatory phenotype [Figure 1].

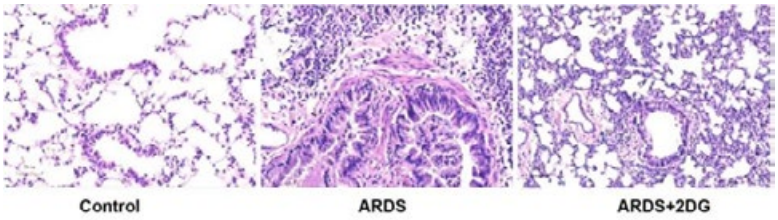

Figure1. In the lung tissue of ARDS mice, there is evident infiltration of inflammatory cells, thickening of alveolar septa and interstitial edema. The glycolysis inhibitor 2-DG can alleviate these pathological changes

Conclusions: Metabolic reprogramming, represented by glycolysis, plays a significant role in alveolar macrophage polarization and modulating the metabolism of alveolar macrophages may be a potential therapeutic approach for ARDS.

Biography

Bo Liu is a cardiothoracic surgeon who has been working at the Children's Hospital of Chongqing Medical University for 7 years. His primary responsibilities include providing diagnosis and treatment for thoracic and cardiovascular-related diseases, training and educating medical students and conducting research on the mechanisms and prevention of acute lung injury. Over the course of his career, he has published numerous research articles in reputable medical journals and has been invited to speak at various international conferences. He is highly respected among his peers and patients alike for his dedication to providing high-quality medical care and his commitment to advancing medical knowledge through research and education.

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Influenza and Pneumonia

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Are symptoms different between Influenza and Pneumonia?

Pneumonia is a lung infection, so it has more respiratory symptoms while influenza is accompanied more by muscular aches and fatigue. Usually pneumonia takes longer to develop and can be a complication of influenza. The flu is caused by a viral infection, while pneumonia can be caused by either a bacterial, viral or fungal infection. Bacterial pneumonia can be an influenza complication. Immunosuppression is a risk factor for the secondary bacterial infection in influenza.

In flu season, every one of all ages is at risk of influenza, but the children are more vulnerable. Every region across the world is susceptible to the contagious respiratory illness, it can range from mild to severe in illness, so many patients with the flu recover within a few days, but some people can develop complications. One of the most significant complications is Pneumonia. One-third of pneumonia cases develop from a respiratory virus, with the flu the most common of those [Figure 1].

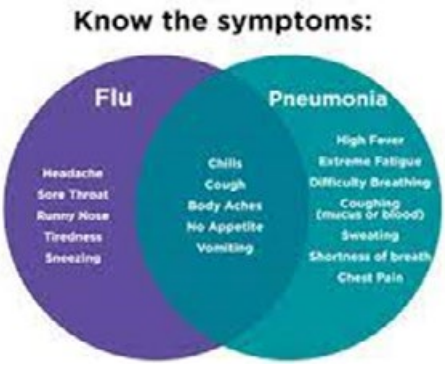

Figure 1. Know the symptoms

Viral pneumonia in healthy people goes away in 1 to 2 weeks, but cough and fatigue may last for many weeks. Viral pneumonia can be serious and life-threatening in people with other medical illnesses. The best thing for a quick recovery from the influenza is rest and hydration, sometimes antiviral medication as oseltamivir can help to avoid complications as Pneumonia, especially in the immunocompromised patients, it can reduce the severity and duration of the symptoms as well.

Simple sanitization by wiping the surfaces with simple detergents, diluted bleach, or hydrogen peroxide, can help stop the spread of the influenza virus and control its seasonal breakdowns. Fortunately, there is a vaccine for both diseases, you can administer a pneumococcal vaccine (PCV15, PCV20 or PPSV23) and also influenza vaccination, this helps a lot to reduce the prevelance of both influenza and pneumonia in the pediatric population.

Biography

Marian Kamal Mankaryous Hendy is a Specialist in the Department of Paediatrics in Medcare Medical Centres. She obtained her MBBS and Master's Degree in Paediatrics and Neonatology from Assiut University in Egypt. She completed both her internship and residency at Assiut University Hospital. She started practicing as a Paediatrician at Assiut University Hospital and the Ministry of Health in Egypt. She then joined the Sohar Hospital in Oman where she worked as a Paediatric Specialist. Subsequently, she also worked as a Paediatric Specialist at Zulekha Hospital in Dubai for 5 years before joining Medcare.

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Congenital syphilis and neonatal cholestasis: A case report

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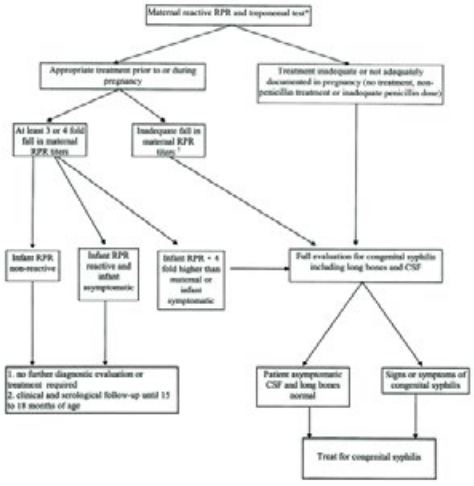
Congenital syphilis remains a disease that leads to a significant number of mortality and morbidity in neonates and new-born's, despite the widespread availability of affordable diagnosis and treatment options for the entire population.

Clinical Case: A new-born, born at full term after 41 weeks of gestation, with a low weight of 2850 kg and an Apgar score of 9/10, was admitted exhibiting refusal to feed, jaundice, and skin desquamation. The mother of the new-born, aged 20 and having had three pregnancies (G3P2), had recently tested negative for syphilis during the third trimester, including a nonreactive VDRL test. Once admitted to the neonatology unit, the new-born underwent a VDRL test, which revealed values of 256 dilutions, along with thrombocytopenia, elevated liver enzymes and hyperbilirubinemia with a predominance of direct bilirubin. A lumbar puncture was performed, and the cerebrospinal fluid (CSF) analysis showed a VDRL value of 2 dilutions, confirming the diagnosis of neurosyphilis. Treatment with penicillin was administered for duration of 10 days. The subsequent course of the disease was complicated by chronic cholestasis during follow-up.

Conclusions: It is crucial to maintain a high level of suspicion for congenital syphilis as a diagnostic approach, even when the mother presents negative test results. This is due to the fact that congenital syphilis remains one of the most prevalent infectious diseases during pregnancy in our environment.



Biography

Silvina Dignani is a doctor in the neonatology service at the Hospital San Felipe de San Nicolás, where she trained and continues to work, having rotated at the Jackson Memorial Hospital in Miami. She was a physician following high-risk premature infants and is currently working in the neonatal ICU service.

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