

3'UTR+132C>T showed normal Hb level, mild microcytic hypochromic, and borderline HbA₂. The same study showed that the co-inheritance of 3'UTR+132C>T and IVS1-1G>A (β^0) was associated with beta-thalassaemia intermedia. Even though this mutation causes a silent or mild phenotype in the heterozygous state, co-inheritance with a significant pathogenic variant of beta mutations will result in a more severe phenotype. In conclusion, the availability of molecular analysis enabled the identification of underlying mutations and improved the understanding of the natural history of the marked clinical heterogeneity of HbE/beta-thalassaemia.

HM40 Prevalence of molecular genotyping of Hereditary Persistence of Foetal Haemoglobin and delta-beta thalassaemia in Malaysia

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Introduction: Hereditary Persistence of Foetal Haemoglobin (HPFH) or delta-beta ($\delta\beta$) thalassaemia causes an increased production of the Hb F level and may worsen the condition if being co-inherited with β -thalassaemia mutation. This study aims to determine the prevalence for common large deletions of β -globin gene clusters associated with high Hb F value among the HPFH or ($\delta\beta$)⁰-thalassaemia carriers in Malaysia. **Materials & Methods:** This retrospective study retrieved 541 cases of HPFH or ($\delta\beta$) thalassaemia carriers diagnosed by Hb analysis from January 2017 until December 2019. Molecular analysis was performed using the multiplex Gap PCR method. **Results:** Among these 541 cases, 63.6% were females and 36.4% were males. For ethnicity, 93.3% were Malay followed by Chinese (5.5%), Indian (0.7%) and others. There were seven subtypes of HPFH and/or $\delta\beta$ -thalassaemia identified. The two most common types of deletion were $G\gamma$ ($\Delta\gamma\delta\beta$)⁰-thalassaemia Siriraj ~118Kb and ($\delta\beta$)⁰-thalassaemia THAI ~12.5Kb with a frequency of 29.9% each. HPFH-6 deletion was detected in 20% of the samples. This is followed by $G\gamma$ ($\Delta\gamma\delta\beta$)⁰-thalassaemia Asian-Indian Inv/Del (14.2%), $G\gamma$ ($\Delta\gamma\delta\beta$)⁰-thalassaemia Chinese ~100Kb (4.4%), HPFH-3 (0.9%) and $G\gamma$ ($\Delta\gamma\delta\beta$)⁰-thalassaemia Asian ~49.3Kb (0.6%). **Discussion:** This study highlights the common HPFH or ($\delta\beta$)⁰-thalassaemia in Malaysia. Identification of these molecular findings may facilitate the diagnostic approach of thalassaemia and haemoglobinopathies in this region.

HM41 Identification of uncharacterised ($\Delta\gamma\delta\beta$) deletion (~101.3Kb) by using multiplex ligation probe-dependent amplification in a Malay family

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Introduction: Hereditary persistence of foetal haemoglobin (HPFH) and delta-beta thalassaemia are heterogeneous conditions caused by a deletion in the beta-globin gene cluster. It is characterised by increased production of foetal haemoglobin (Hb F) levels in adulthood. **Case report:** A five-year-old Malay girl and her parents' samples were referred to Institute for Medical Research for beta thalassaemia genotyping. The haemoglobin value of the index patient was 12.9 g/dL with MCV and MCH values of 67.7fL and 21.9pg, respectively. Her Hb analysis findings revealed a normal Hb A₂ of 2.6% with an increased level of Hb F (24%) leading to a presumptive diagnosis of delta-beta thalassaemia or HPFH trait. Common beta-globin gene cluster deletions were ruled out using the Multiplex Gap method. Further investigation was done using Multiplex Ligation-dependent Probe Amplification (MLPA) assay. **Discussion:** No common deletion was found using the Multiplex Gap method. MLPA findings of the index patient and her father revealed a heterozygous state of the uncharacterised deletion spanning from upstream of *HBG1* until *OR51V1* gene (downstream of *HBB*). Based on the MLPA probe to probe distance, the estimated size of the deletion is about ~101.3Kb which involves *HBG1*, *HBD* and *HBB* genes while leaving the *HBG2* intact. This uncharacterised deletion leads to a new ($\Delta\gamma\delta\beta$) deletion being identified. However further investigation is essential to characterise the deletion breakpoint in order to determine the actual size and further understanding of the deletion. A combination of this uncharacterised deletion with other beta thalassaemia mutation/deletion may result in thalassaemia intermedia.

HM42 Acute lower limb ischemia presenting Acute Myeloid Leukaemia

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Introduction: Patients with underlying acute myeloid leukaemia (AML) may develop disorders of the coagulation system, leading to the more commonly seen haemorrhagic complications. Thrombosis in leukaemia occurs more commonly in veins and rarely in arteries. We report a case of a middle-aged lady who presented with an acute lower limb ischemia as

the initial manifestation of undiagnosed acute leukaemia. *Case Report:* A 57-year-old lady presented with sudden onset, unprovoked left lower limb pain. There was no preceding trauma or infection. She had mild, intermittent episodes of gum bleeding in the preceding three weeks. Posterior tibial and dorsalis pedis pulses of her lower limb were absent with ankle: brachial pressure index less than 0.5. Physical examination was unremarkable. A CT angiography showed presence of a long segment thrombosis involving the left common and external iliac arteries. 84% blast cells with leucoerythroblastic picture were seen on PBF. Bone marrow aspirate demonstrated 77% blasts which were positive for peroxidase, suggestive of AML. *Discussion:* Thrombotic events are less commonly seen in AML, due to thrombocytopenia accompanying coagulopathy in affected patients. Thrombosis usually occurs in small vessels; large artery thrombosis is rarely seen. This case illustrates one of the rare instances where a patient with acute myeloid leukaemia developed large lower limb artery thrombosis, leading to unilateral acute limb ischaemia. It is important to always keep in mind the possibility of haematological malignancies as a cause of acute limb ischaemia, albeit rare, so as to institute prompt and appropriate therapy for the patient.

MEDICAL MICROBIOLOGY

MM01 Community-Associated Methicillin-Resistant *Staphylococcus aureus* (CA-MRSA): antimicrobial susceptibility patterns versus *mecA* gene and Panton-Valentine Leukocidin (PVL) genes in clinical isolates from Hospital Tuanku Ja'afar, Seremban

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Introduction: The emergence of CA-MRSA is well recognised as a significant pathogen in the public and healthcare-associated settings. The aim of this study was to determine the prevalence of CA-MRSA carrying *mecA* (*SCCmecA*) and Panton-Valentine leukocidin (PVL) genes in clinical isolates from Hospital Tuanku Ja'afar. *Materials & Methods:* All non-duplicate clinical isolates tested for MRSA with antimicrobial susceptibility patterns showing a possibility of community-acquired MRSA, which was resistance only to cloxacillin and penicillin disks by the diffusion method were prospectively collected at Hospital Tuanku Ja'afar from July 2017 to June 2018. The isolates were sent to the Institute of Medical Research for genotyping of the *mecA* and PVL genes. *Results:* A total of 58 non-repeated isolates, which were phenotypically positive for CA-MRSA, were recovered. Among them, 40 isolates were subjected to *mecA* and PVL gene detection. All isolates were clindamycin susceptible. Of the 40 isolates, 22 (55%) carried staphylococcal cassette chromosome *mecA* (*SCCmecA*) and Panton-Valentine leukocidin (PVL) genes. 18 (45%) were detected with *mecA* but not with PVL. *Discussion:* Based on our study, we can conclude that the phenotypic characteristic of CA-MRSA can be used as a screening method as 55% of clinical isolate in our centre produce the PVL gene. Further study needs to be done, and this is the limitation of our research as we were unable to determine the sequence type (ST).

MM02 *Bordetella trematum* bacteremia in a leukaemic patient and review of previous cases

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Introduction: *Bordetella trematum* is a gram-negative coccobacilli and was first reported in 1996. It is a relatively rare organism causing infection rendering information on the pathogenesis, life cycle and virulence limited. There is also no standardised method and interpretive criteria for antimicrobial susceptibility testing for this organism. When reported, this organism is highly associated with immunocompromised and diabetic patients. *Case report:* This is a case of a newly diagnosed leukaemic patient which first presented with a history of unresolving respiratory infection despite receiving multiple courses of antibiotics. *Bordetella trematum* was isolated from blood culture with identification using VITEK 2 GN (Gram-negative bacilli). After a course of ceftriaxone followed by piperacillin-tazobactam, the patient improved clinically and was discharged well. *Discussion:* Majority of isolated *Bordetella trematum* were from tissue and swab samples of infected wounds with polymicrobial infection while isolation from blood have been infrequent. Although still rare, it is increasingly reported, as laboratories gain greater access to technologies for accurate and specific bacterial identification. As it may be an emerging microorganism, monitoring and reporting the isolation of this organism is essential to add knowledge on the clinical significance and antimicrobial susceptibility pattern.

MM03 Carbapenem heteroresistance in colistin non-susceptible *Enterobacter asburiae*: a case report

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Introduction: The emergence of colistin resistance in the *Enterobacter cloacae* complex, particularly *Enterobacter asburiae* is alarming, especially in carbapenem-resistant isolate. This case highlights the importance of laboratory detection of