27th August 2014 (Wednesday)
1445 - 1645
Selangor Room

YOUNG INVESTIGATOR AWARD PRESENTATIONS

Chairperson: *Chooi-Fun Leong*

YIA-OP01. Thalassaemia screening in newborns using dried cord blood spots: a preliminary observation

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Introduction: Beta-Thalassaemia major and intermedia patients suffer from complications related to transfusion-dependent anemia while alpha thalassaemia major or hydrops foetalis is incompatible with life. The prevalence of thalassaemia carriers in Malaysia ranges from 10-15%. Most people are oblivious as there is no standard screening program in place. *Objective:* To compare Haemoglobin (Hb) analysis using cord blood whole blood (CBWB) and dried cord blood spot (DCBS). *Materials and method:* Twenty samples from term neonates were collected; 3ml CBWB samples in K2EDTA and DCBS on whatman filter paper. Hb analysis was performed at Red Cell Unit, Hospital Ampang using the Neonate program on Sebia Capillary's 2 automated systems. Kleihauer test was performed to exclude contamination by maternal blood in the CBWB. The HbA and HbF percentages were compared and HbA:F ratio was calculated. The data was analyzed on SPSS version 22. *Results:* Five samples showed presence of variant Hb for both sampling methods; two are "suggestive of HbE trait", 1 sample each is "suggestive of HbS trait", "suggestive of HbBart's trait" and "suggestive of HbD-Punjab trait". In CBWB, the mean HbA was 20.3% and HbF was 78.4%. In DCBS, mean Hb A was 12.27% and HbF level was 47.4%. Correlation coefficient between CBWB and DCBS showed r = 0.792 for HbA and 0.771 for HbF. In 2 samples, HbA:F ratio showed ≤ 0.1, which may be suspected for Beta-thalassaemia trait. The HbA:F ratio between the two samples showed r = 0.676. Kleihauer test was negative for all samples. *Discussion and Conclusion:* No maternal contamination was detected in all cord blood samples. The comparative data shows Hb fractions analyzed on CBWB and DCBS is almost similar, demonstrating good correlation. However, all results must be confirmed with molecular analysis. Proper techniques should be established to avoid maternal contamination in cord blood samples. DCBS offers a non-invasive collection method and high stability at room temperature, suitable for areas of low resource settings. Neonatal thalassaemia screening using DCBS could offer a first line detection of thalassaemia carriers in Malaysia.

YIA-OP02. The association of Ki67 score with type and grading of cervical adenocarcinoma: an experience with 41 cases

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Introduction: The incidence of adenocarcinoma of the cervix is increasing worldwide. It accounts for up to 25% of all cervical carcinomas. The Ki 67 has shown some potential in prognostication in

cancers; however their uses in cervical cancers have not been explored. Thus, this research aims to address the significance of Ki 67 expression in adenocarcinoma of cervix and its association with histological type and grading on cervical adenocarcinoma. *Materials and methods:* Paraffinized archival tissue blocks of cervical adenocarcinoma in year 2006 to 2010 from Histopathology Unit, Department Pathology, Hospital Kuala Lumpur was retrieved. The blocks were sectioned and stained for Hematoxylin and Eosin (H&E) and immunohistochemistry. The H&E sections were reviewed for confirmation of diagnosis, histologic type and grading before the immunohistochemical stains were done. The positive Ki-67 expression stained the nuclei brown in immunostained sections. The score of Ki-67 was then determined. A cut off level of 30% was used to define high and low proliferating tumour. *Results:* Total of 69 cases was retrieved. However, after exclusion, only 41 cases were used in the study. The youngest patient was aged 23 years old while the oldest patient included in this study was 71 years old. The mean age of the patients was 48.61 ± 11.33 (mean ± SD) years old. Most of the cases were mucinous adenocarcinoma (68.3%), followed by NOS (19.5%), endometrioid (7.3%) and serous adenocarcinomas (4.9%). The mucinous cervical adenocarcinoma was predominantly endocervical subtype. Majority (46.3%) of cervical adenocarcinomas were of high grade (moderate and poor). The mean value of Ki 67 index was 38.15 ± 16.49% (mean ± SD) with the majority of adecarcinomas having an index between 41% to 50%. However, there was no significant association seen between the Ki 67 index with histological type or grading. *Conclusion:* Adenocarcinoma of cervix was mainly diagnosed in women in their 40s. The Ki 67 score is more than 30% in most adenocarcinoma. There was no statistically significant association between the Ki 67 index with histological type or grading of cervical adenocarcinoma.

YIA-OP03. Low DNA concentration from buccal swab can be used for blood group molecular genotyping

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Introduction: Blood has been the sample of choice for DNA analysis in many conditions. However, blood sampling is invasive, time consuming and sometimes difficult to obtain. In repeatedly transfused patients, accurate red cell antigen typing by serology is a constant problem due to exposure to blood donors' DNA. To overcome these problems, another source for DNA analysis is needed. *Materials & methods:* 12 buccal swab scrappings were collected using sterile cotton wool swab from healthy volunteers. The DNA concentration yeild was below 10 ng/µl. 3.5ml of peripheral blood sample was also collected for serological red cell phenotyping. DNA was extracted using a commercial kit (QIAamp DNA Blood Mini Kit, Qiagen). SNP RT-PCR was performed using the oligonucleotide primers for RHE, FY^a/FY^b and JK^a/JK^b (rs609320, rs1058396 and rs12075, respectively) and Taqman probes were designed and synthesized by Applied Biosystems. The fluorescence data was analyzed with allelic discrimination 7500 Software, v.2.0.2. Genotyping results were compared to the result of serological blood grouping using standard hemagglutination techniques. *Results:* Genotyping results from buccal swab samples and phenotyping results via serology were concordant for all samples tested. Two volunteers were heterozygous Rh E/e and 10 volunteers were homozygous Rh e/e. For Duffy blood group antigen, 9 volunteers were Fy(a+b-), 2 volunteers were Fy(a+b+) and 1 volunteer was Fy(a-b+). For Kidd blood group antigen, 4 volunteers were Jk(a+b-), Jk(a+b+) and Jk(a-b+) each. *Discussion & Conclusion:* Buccal swab samples that yield low DNA concentration can still be used for accurate blood group genotyping. This is of particular value in genotyping repeatedly transfused patients or patients whose red blood cells are coated with antibodies that renders the serological method for genotyping and phenotyping invalid. Buccal swab to collect epithelial cells offers a simple and inexpensive alternative collection method ideal for whole-genome amplification.

YIA-OP04. Vascular malformations and neoplasms: an effort to reclassify the commonest soft tissue lesion in children and adolescents

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Introduction: Vascular anomalies are the commonest soft tissue lesions in children and adolescents. This group of lesions has seen significant advances in diagnosis, new therapies and genetic understandings in recent years. The latest change in the classification systems warrants an effort to reclassify our cases to streamline the diagnosis which could affect therapeutic modalities. *Methods and materials:* We performed a retrospective review of all paediatric and adolescent cases in Hospital Kuala Lumpur from year 2011 – 2013. In our attempt to reclassify these cases, we applied specific histological criteria and support our findings by use of immunohistochemical markers, GLUT-1 and Podoplanin. *Results:* 60 cases were reviewed and reassessed; of which 46 cases were vascular malformations, and 9 cases were vascular neoplasms. Another 5 cases were called reactive vascular proliferation. Of the 46 cases, there were 26 lymphatic malformations, 6 cavernous hemangiomas, 6 capillary malformations, 5 venous malformations, 1 combined AV malformations, 1 Masson's tumour, and a child with both lymphatic malformations and Masson's tumour. The 9 cases of vascular neoplasms include 4 infantile hemangiomas (IH), 3 tufted angiomas, and 2 Kaposiform hemangioendotheliomas. All 5 cases of reactive vascular proliferation were lobular capillary hemangiomas. *Conclusion:* The use of GLUT-1 has helped in the diagnosis and differentiation of IH from capillary hemangioma, lobular capillary hemangioma and Kaposiform hemangioendothelioma. Although our study involved a small group of patients, it revealed IH as the commonest benign vascular tumour, affecting 40% of all the neoplastic case and correlating well with international data. We also found that all 4 patients with IH were female infants. There is a need to identify IH as this benign tumour most often do not require surgical or medical interventions, unlike the other tumours which require either surgical treatment, immunotherapy such as Interferon or pharmacotherapy such as Vincristine.