receptor and/or progesterone receptor in immature female rat and GH3 cell line. An increasing number of chemical compounds in the environment have been identified as endocrine disruptor in vivo and in vitro bioassay. A future challenge is required to confirm a theoretical toxicology and risk assessment of EDs for human and animal health.

**Keywords:** Endocrine disruptors (EDs); theoretical toxicology; risk assessment; reproductive system

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### AB160. Fuminal hepatic failure in Wilson disease

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**Background:** Wilson disease (WD) is an autosomal recessively inherited disorder of copper metabolism. Mutation of the ATP7B gene on chromosome 13 leads to accumulation of copper in the liver, brain, kidney and cornea. Clinical presentation is particularly in liver and central nervous system. Fuminal hepatic failure is rare and mortality rate is very high.

**Methods:** Retrospective description. Describe clinical, preclinical characteristics of patients acute liver failure due to WD.

**Results:** A total of 6 patients with acute liver failure due to WD (Leipzig 2001) at Hepatology Department in NHP from January 2014 to June 2015. Common symptoms: jaundice, edema, ascites, hepatic encephalopathy, hyperbilirubinemia, hemolytic anemia, hypoalbuminemia, severe coagulation disorder, ceruloplasmin <0.2 g/L, urinary copper/24 h >100 mcg/dL. Five patients improved after treatment, one patient died due to fulminant liver failure.

**Conclusions:** Fulminant hepatic failure which cause by WD is very rare in children. The disease can be rescued if it is diagnosed and treated promptly. WD should be suspected in fuminal hepatic failure unknown the origin.

**Keywords:** Wilson disease (WD); fuminal hepatic failure

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### AB161. High resolution melting analysis of buccal DNA revealed a significant association between UGT1A1 c.211G>A and neonatal hyperbilirubinemia development in Malay population

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**Background:** Severe neonatal hyperbilirubinemia or neonatal jaundice (NNJ) characterised by an elevated total serum bilirubin (TSB) level may result in kernicterus or even death. Uridine diphosphate glucuronosyltransferase 1A1 (UGT1A1) is the key enzyme which conjugates bilirubin with glucuronic acid for the subsequent bilirubin excretion. Conversely, constitutive androstane receptor (CAR), encoded by nuclear receptor subfamily 1, group I, member 3 (NR1I3) gene, regulates bilirubin excretion by activating the components of the bilirubin clearance pathway. Thus, genetic variants in UGT1A1 and NR1I3 genes may modulate bilirubin excretion and lead to NNJ. This study aimed to determine the association between UGT1A1 and
NR1I3 genetic variants and NNJ development in Malay population by genotyping the DNA isolated from buccal swabs. The accuracy and reliability of the genotyping results produced by buccal DNA was also compared with that of the whole blood DNA.

Methods: Buccal swabs were collected from 232 hyperbilirubinemia and 232 non-hyperbilirubinemia newborns admitted to and/or born in Hospital Universiti Sains Malaysia (HUSM). Hyperbilirubinemia subjects were those with TSB levels ≥250 µmol/L within the first week after birth while non-hyperbilirubinemia subjects were newborns without significant hyperbilirubinemia. The UGT1A1 (c.211G>A) and NR1I3 [MPJ6_1I3008 (G>A), IVS8+116T>G and 540A>G] variants were genotyped by using high resolution melting (HRM) analysis. Binary logistic regression was used to assess the association between variant genotypes and risk of NNJ. Whole blood samples were also collected from 60 subjects and genotyped to compare the HRM genotyping results with that of the buccal swabs.

Results: When compared with wild-type genotype, both heterozygous and homozygous variant genotypes of MPJ6_1I3008 (G>A), IVS8+116T>G and 540A>G were not significantly associated with NNJ. However, the heterozygous genotype (GA) of c.211G>A was found to increase the risk of NNJ (OR: 1.96, 95% CI, 1.13-3.39, P=0.014). Besides, all buccal DNA samples demonstrated 100% genotype call rates and achieved complete genotype concordance with blood DNA samples.

Conclusions: The heterozygous genotype of c.211G>A could be a genetic risk factor of NNJ in Malay population. Since buccal DNA produced complete genotype call and concordance rates, the non-invasive buccal swabs collection can be used as an alternative to blood sampling especially in genetic studies involving paediatric population.

Keywords: Neonatal hyperbilirubinemia; UGT1A1; NR1I3; buccal cells; Malay


AB162. Genes variation in three families of Vietnamese dioxin victim

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Abstract: Dioxins are a class of chemical contaminants that are formed during combustion processes such as herbicide manufacturing, waste incineration, forest fires, and backyard trash burning. The most toxic chemical in the class is 2,3,7,8-tetrachlorodibenzo(para-dioxin (TCDD). Approximately 18 million gallons of Agent Orange were sprayed by US Airforce on southern of Vietnam from 1962 to 1971. About 0.3% of Agent Orange consisted of TCDD. Dioxins have been considered highly toxic and able to cause cancer, reproductive and developmental problems, damage the immune system, and interfere with hormones. In this paper we studied gene variation in some families of dioxin victims of Vietnamese army veterans who have been exposed directly under sprays or carried out missions for at least 2 years in the heavily sprayed regions. Of the first family, we found 21 nucleotide variants in TP53 gene, 13 nucleotide variants in AhR gene. All of them leading to amino acid change. We also found R554K in ThB4.VT16 and ThB4.VT17. This mutation changes activity for CYP1A1 induction in lymphocytes. In the second family, we identified 29 nucleotide variants in TP53 gene. Although we could not found any variant associated with phenotype of the family members but previous studies have found P295L associated with gastric carcinoma, L299P associated with pancreatic cancer, G279E associated with colorectal carcinoma and cancer of male sex cells. In the third family, we found 22 nucleotide variants in TP53 gene and 9 variants in CYP1B1 gene. For understanding of whole genome sequence variation, whole genome of 3 member of each family has been sequenced by Illumina HiSeq 2000/2500 platform. The whole genome sequence data have started analysing.

Keywords: AhR; CYP1A1; dioxin; TP53; whole genome