Validation and clinical case report in non-invasive prenatal testing for all chromosomes

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ABSTRACT

Introduction: Whole-Genome Sequencing (WGS)-based bioinformatics algorithms offer the potential to detect aneuploidies for all chromosomes. However, current non-invasive prenatal testing (NIPT) mainly focuses on chromosome 21, 18, 13, and sex chromosomes, leaving the need for a more accurate detection algorithm for Rare Autosomal Trisomies (RATs) that can provide insights into feto-placental biology.

Methods: To address this, we conducted a literature search to identify RAT cases and created artificial data for each case based on different fetal fractions. Using our in-house pipeline, we applied a z-score analysis twice to maximize sensitivity and set the threshold. Additionally, we calculated demographic statistics using clinical data.

Results: Our results showed varying accuracy for each chromosome depending on the fetal fraction. With a fetal fraction of 10%, the proposed algorithm achieved high accuracy (>99%) for most chromosomes, and even at a fetal fraction of 5%, most chromosomes had accuracy above 95%. However, chromosome 19 displayed lower accuracy at 80%. Out of 30,364 clinical samples, we identified 187 RATs, with Trisomy 7 (n=58) being the most frequent, followed by Trisomy 16 (n=17) and Trisomy 8 (n=16). Of the 23 cases that underwent amniotic karyotyping, 6 confirmed abnormalities (mosaic or trisomy), while the other 17 cases showed normal results, suggesting confined placental mosaicism.

Conclusions: Our algorithm, supported by well-designed artificial samples, demonstrates the ability to detect aneuploidies for all chromosomes. Comprehensive chromosome testing can provide valuable information on the presence of RATs, which may impact pregnancy outcomes through placental dysfunction, fetal growth restriction, and potential uniparental disomy.

An overview of postpartum haemorrhage (PPH) incidence and risk factors in Kepala Batas Hospital, Penang from year 2020 to 2022: A retrospective study

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ABSTRACT

Introduction: Ministry of Health Malaysia recorded 26% maternal morbidity due to postpartum haemorrhage (PPH) in the year 2017. This retrospective study was aimed to determine the incidence, associated risks factors, management, and perinatal outcomes of postpartum haemorrhage in our centre. Methods: We performed a retrospective study, which included all the PPH cases delivered in Hospital Kepala Batas (HKB), Penang from 1st January 2020 to 31st December 2022. Clinical data and patients’ demographics were collected and analysed using SPSS version 21. Results: The incidence of PPH in HKB was 3.6%. The commonest risk factors among primary PPH patients were augmentation of labor (41.8 %), followed by anemia in pregnancy (37.3 %), and induction of labour with prostin (21.8%). The leading co-morbidity among PPH patients was diabetes (32.7 %). The major cause of PPH is perineal tear (50.8%) and uterine atony (47.3 %). The most common approach in the management of PPH are blood transfusion (41.8 %), intravenous injection of hemabate (39.1 %), repeat syntometrine (19.1%), and parenteral iron, hemfero (15.5%). There were significant associations between estimated blood loss, and 1) patients age, 2) fetal distress case, 3) augmentation of labour, 4) delivery mode, 5) cause of PPH (intrapartum risk, tear), and 6) intervention (blood transfusion, uterogenic agents). All the patients survived. Conclusions: The incidence of PPH in HKB is lower than that of population-based studies (5%). The timely intervention resulted in no maternal mortality.