ISP-17-6  Uniparental disomy analysis in trios using genome-wide SNP array and whole-genome sequencing data imply the segmental uniparental isodisomy in general populations

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[Aim] It is known that a whole chromosomal and segmental uniparental disomy (UPD) is one of the causes of imprinting disorder and other recessive disorders. The aim of this study was to investigate a whole chromosomal and segmental UPD in general population. [Materials and Methods] All samples were obtained after receiving written informed consent and the study protocol was approved by the IRB. Here, we present results of a whole chromosome and segmental UPD analysis using single nucleotide polymorphism (SNP) microarray data of 173 mother–father–child trios (519 individuals) from six populations (including 170 HapMap trios). [Results] We identified obvious one segmental paternal uniparental isodisomy (UPD) (8.2 mega bases (Mb)) in one HapMap sample from 173 trios using Genome-Wide Human SNP Array 6.0 (SNP6.0 array) data. On the other hand, we could not find the shorter segmental iUPD in two trios using whole-genome sequencing data. Finally, we estimated the rate of segmental UPD to be one per 173 births (0.578%) based on the UPD screening for 173 trios in general populations. Based on investigated autosomal chromosome pairs, we estimate the rate of segmental UPD to be one per 3806 chromosome pairs (0.026%). [Conclusion] These data will imply the possibility of hidden segmental UPD in normal individuals.

ISP-17-7  Monitoring of intrinsic optical signals relating to cerebral hemodynamics and cellular morphology in a rat hypoxic ischemic encephalopathy model

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[Objective] For hypoxic ischemic encephalopathy (HIE) during labor, no valid diagnosis is established. In this study, we aimed to identify intrinsic optical signals to detect tissue deterioration prior to the occurrence of HIE during labor using a rat model. [Methods] Seven days old rats were underwent left carotid artery ligation, and 120 minutes later the rats were exposed to 8% oxygen for 135 minutes. Time courses of diffuse reflectance intensities were measured to monitor total hemoglobin, deoxy-Hb concentration and light scattering signal associated with cellular morphology. Staining for mitochondria by triphenyltetrazolium chloride (TTC) was conducted to evaluate brain tissue deterioration caused by HIE. [Results] During hypoxia, the diffuse reflectance intensities corresponding to total hemoglobin and deoxy-Hb concentration indicated the reduction in cerebral blood flow and deoxygenation of blood in the ligation hemisphere were more remarkable in the ligation hemisphere than in the non-ligation hemisphere. Light scattering signal was reduced in the ligation hemisphere, which may indicate edema formation. In agreement with the data of optical signals, TTC staining showed avascular necrosis only in the ligation hemisphere. [Conclusion] The optical signals measured in this study are promising parameters to monitor brain tissue deterioration triggered by hypoxia during labor.

ISP-17-8  Prenatal diagnosis of urorectal septum malformation sequence with the description of new variant. Insight into the embryogenesis

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[Objectives] Urorectal septum malformation (URSM) sequence is a spectrum of anomalies that includes the absence of urethral and vaginal, external genital defects and anorectal abnormalities. We reviewed 4 cases in a single center, Taiwan University Hospital, over a period of 20 years (1995–2015) to evaluate possible embryogenesis and histology of URSM. [Methods] Four cases diagnosed with URSM prenatally were included in this study. We collected the data of clinical presentations, prenatal ultrasonography figures, and final autopsy results and reviewed the current literatures. [Results] All of our cases had the similar clinical features, such as ambiguous genitalia, imperforate anus, invisible perineal opening or urethral opening. Pelvic cysts, oligohydramnios, urinary bladder agenesis, and renal dysplasia were noted during the prenatal examination in three cases. A new variant as intermediate form of URSM was noticed in case1; because that we were unable to locate it in this full continuum of spectrum due to the URS has formed but there is no perineal opening. Therefore, a preferable name such as "URS (representing urorectal septum) – CM (representing cloaca membrane) malformation sequence" might be more appropriate to describe the pathogenesis and manifestation of this whole disease spectrum. [Conclusion] Faulty development of URS apparently cannot explain the whole spectrum of the disease. We strongly recommended using URS-CM malformation to describe the original spectrum of URSM. More clinical cases and animal studies should be collected to verify this notion.