

## AB034. Carrier frequency of inherited genetic disorders in Thai population: implication for designing expanded carrier screening panel

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**Background:** Expanded carrier screening (ECS) is the process of screening individuals for hundreds of autosomal recessive (AR) and X-linked recessive (XLR) diseases. ECS result is beneficial for couples who want to make an informed reproductive choice. Though next-generation sequencing allows thousands of genes to be sequenced at once, the ideal ECS gene panel should only include diseases that its carriers are common in the population (1% or higher), and the prenatal diagnosis of the disease is allowed. However, designing the ECS panel for Thais can be difficult because the carrier frequency for most genetic diseases is unknown. Therefore, we retrospectively review our exome sequencing data from Thai patient to get an estimation of the carrier frequency.

**Methods:** Variants in 540 genes associated with AR and

XLR from exome sequencing data of 178 unrelated Thai patients are filtered and matched against known pathogenic mutations in reputable mutation databases (OMIM, Clinvar, and HGMD).

**Results:** Pathogenic mutations in *SLC26A4* gene associated with Pendred syndrome have the highest carrier frequency of 2.81%. Usher syndrome (*USH2A*) is the second-most common with the carrier frequency of 2.25%. 1.69% of the patients also have pathogenic mutations in the gene related to impaired vision and impaired hearing such as *OCA2*, *OTOF*, *CLCN1* *BSND* and *AHI1*. Only 1.12% or less is the carrier of severe diseases such as myotonia congenita (*CLCN1*), lactic acidosis (*POLG*) and molybdenum cofactor deficiency (*MOCSI*).

**Conclusions:** Inherited diseases that involved the impairment of hearing and vision are somewhat common among Thais. The ethical issue could arise when these diseases are included the ECS panel because they are not severe enough to allow for prenatal diagnosis. More discussion about ethics of ECS and more data on carrier frequency are needed to offer ECS ethically.

**Keywords:** Expanded carrier screening (ECS); ethical issue; next generation sequencing; carrier frequency; Thai population

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