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Dec 17, 2020

Dear Doctors,

We deeply appreciate your support and trust in our services. We are writing this letter to update you that ***CYP21A2*** gene for **Congenital adrenal hyperplasia due to 21-hydroxylase deficiency (21-OH CAH)** has been added to Invitae Comprehensive Carrier Screening panel, which is effective immediately **17-Dec-20.** Therefore, Comprehensive Carrier Screening panel will automatically include the *CYP21A2* gene and in total will be **302** genes. Please be reminded that due to the limitations of next generation sequencing, currently Invitae only analyzes the most common mutations and rare selected HGMD variants of *CYP21A2* gene, and duplications are reported only in the presence of a pathogenic variant(s). **The detection rate of *CYP21A2* gene by this panel is 92%**. For more details, please kindly visit *Assay and technical information* on Invitae website (https://www.invitae.com/en/physician/tests/60100/#info-panel-assay_information).

21-OH CAH is an **autosomal recessive** rare disorder involving impaired synthesis of cortisol from cholesterol by the adrenal cortex(https://www.ncbi.nlm.nih.gov/books/NBK1171). It includes the salt-wasting form, simple virilizing form and non-classic form. Newborns with salt-wasting 21-OHD CAH are at risk for life-threatening salt-wasting crises. Most affected individuals are compound heterozygotes, which are inherited from their parents. According to our published data (*HKMJ* in-press), the carrier rate of 21-OH CAH in **Hong Kong Chinese population is 7.7% (1 in 13)**. Therefore, including this gene in carrier screening would provide additional assessment of reproductive risk to support an informed fertility decision.

Please feel free to contact the Prenatal Genetic Diagnosis Centre, Department of Obstetrics and Gynaecology if you have any further questions.

On behalf of Dr. Richard CHOY
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