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CAH
Autosomal recessive

Molecular Genetics Service Profile **CYP21 (CYP21A2, P450c21)**

Introduction

- ✓ The CYP21 gene has been mapped to chromosome 6p21.3, spans 3.34 kb, and consists of 10 exons, which encode a predicted protein of 495 amino acids.
- ✓ The 21-hydroxylase pseudogene (CYP21P) is situated on 6p, close to CYP21, which predisposes the functional gene to mutation through gene conversion or through deletion by homologous recombination and unequal crossing-over.
- ✓ Mutations within the CYP21 gene lead to autosomal recessively inherited **21-hydroxylase deficiency** causing more than 90 percent of cases with **Congenital Adrenal Hyperplasia (CAH)**.
- ✓ A severe form with a concurrent defect in aldosterone biosynthesis (salt-wasting type) and a form with apparently normal aldosterone biosynthesis (simple virilizing type) are together termed classic 21-hydroxylase deficiency. There is also a mild, nonclassic form that may be asymptomatic or associated with signs of postnatal androgen excess.
- ✓ Patients may present with clinical features such as **salt wasting, ambiguous genitalia, postnatal virilization, accelerated growth rate, oligomenorrhea**.

Please photocopy and distribute this sheet as required

Reasons for referral

- ✓ Mutation screening in patients with clinically confirmed or suspected CAH.

Samples

- ✓ Minimum of 2 ml blood sample in EDTA (or minimum of 50 µg DNA from peripheral lymphocytes) can be sent to our laboratory by express mail. In special cases a investigation of DNA from prenatal samples can be made, however you should contact our laboratory for further details.

Technical

- ✓ Mutation scanning of exons 1-10 of CYP21 by dHPLC (WAVE), denaturing high pressure liquid chromatography. Fragments with abnormal elution patterns are directly analyzed with Dideoxy sequencing (ABI 310).

Target turn-round time

- ✓ 3-4 weeks from the receipt of all required samples and clinical information.

Cost

- ✓ CAH Full mutation screen - € 800. In special cases we will provide this service as part of our research program. Please contact us directly.

References

- ✓ Prader A "Der Genitalbefund beim Pseudohermaphroditismus femininus des kongenitalen adrenogenitalen Syndroms" Helv Paediatr Acta. 1954 Jul;9(3):231-48.
- ✓ Speiser PW and White PC "Congenital adrenal hyperplasia. Review." N Engl J Med. 2003 Aug 21;349(8):776-88.

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