17α-Hydroxyprogesterone

Description	Intermediate in the adrenal steroid synthetic pathway.
Indication	Diagnosis and monitoring of congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency.
Additional Info	21-hydroxylase is required for adrenal synthesis of cortisol and aldosterone. In 21-hydroxylase deficiency, metabolic intermediates proximal to the block, including 17-hydroxy-progesterone, accumulate and are shunted into androgen synthesis. Cortisol deficiency leads to loss of negative feedback on ACTH, resulting in increased steroidogenesis proximal to the enzyme block and hyperplasia of the adrenal cortex. Three forms of 21-hydroxylase deficiency CAH have been defined. (1) Severe classical salt-wasting: elevated androgens cause ambiguous genitalia in the affected female foetus, and aldosterone synthesis is impaired. (2) Classical simple-virilising: aldosterone synthesis is normal, but affected females present with genital ambiguity. (3) Nonclassical: aldosterone synthesis is normal and affected females do not have genital ambiguity, but may present with signs of hyperandrogenisation in childhood or adulthood.
Concurrent Tests	Testosterone, DHEAS, androstenedione (all suspected CAH) ACTH, cortisol (suspected classical CAH only)
Dietary Requirements	N/A
Interpretation	Full term babies > 48 hours old: Reference range <20 nmol/L. Premature and sick babies may show moderately increased levels. High levels are found in 21-hydroxylase deficiency CAH; tend to be unequivocally raised (> 150 nmol/L). Adults: reference range <8 nmol/L. In CAH heterozygotes and mild late-onset forms there is a greater than normal rise in 17α-hydroxyprogesterone following ACTH injection (>30nmol/L).
Collection Conditions	Samples for diagnosis of 21-hydroxylase deficiency should be taken at least 48 hours postpartum and preferably before any steroid therapy. Contact laboratory (Duty Endocrine Biochemist) before sending sample if request is an emergency investigation. In the investigation of late onset (or mild) CAH, a random sample is recommended initially, but levels can also be measured at baseline, 30 and 60 minutes following stimulation with Synacthen.
Frequency of testing	Minimum retesting interval in female androgen excess – 1 year.

	Samples may be required for monitoring of steroid replacement therapy in confirmed CAH. Blood spots dried onto filter paper may be a more convenient sample type – this should be pre-arranged with laboratory. (Blood spots must NOT be used for screening/diagnosis.)