Bloodspot 17-hydroxyprogesterone; 17-OHP

Monitoring of patients with Congenital Adrenal Hyperplasia (CAH)

Pseudonyms: 17α-hydroxyprogesterone (17α-OHP), 17α-hydroxypregn-4-ene-3,20-dione

General information

Collection Container: Bloodspot card

Type and volume of sample: 2 bloodspot samples should be collected at 3 different times throughout one day (6 bloodspots in total)

Specimen transport/special precautions: Patients are instructed to collect free flowing blood from a finger prick onto Perkin Elmer 226 filter paper and note the date and time of collection.

Laboratory information

Method principle: 17α-OHP is analysed using a liquid chromatography-high resolution accurate mass- mass spectrometer (LC-HRAM-MS).

Biological reference range: N/A – for monitoring purposes only

Turnaround time: 1 month

Clinical information

There are various congenital enzyme defects of the steroid biosynthesis which cause congenital adrenal hyperplasia (CAH). They are genetically different, but are all transmitted in an autosomal recessive mode. The most frequent types are the 21-hydroxylase deficiency (>90% of cases) and the 11β-hydroxylase deficiency (approx 5-8% of all cases). 17α-OH-progesterone, a precursor of cortisol, is increased in both 21- and 11β-hydroxylase deficiency, but not in other types (see figure 1).

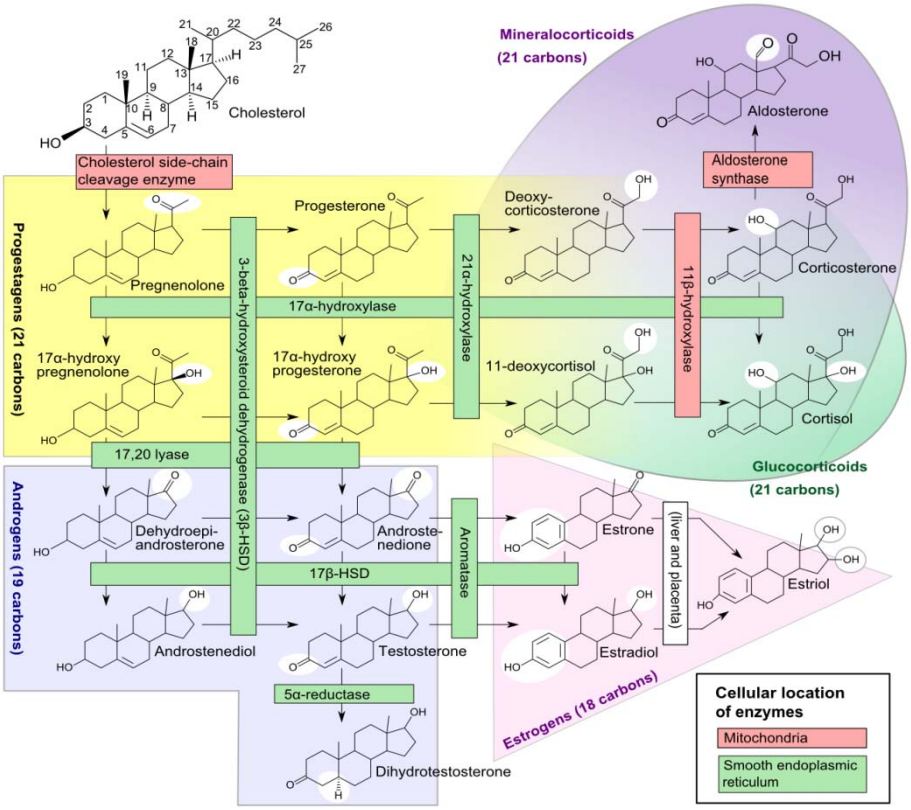

Figure 1: Steroidogenesis. The enzymes affected in CAH are represented by one red and four green bars on the top half of the diagram

The assay has been found to have no cross-reactivity with the following compounds: DHEAS, androstenedione, testosterone, DHEA, epi-Testo, cortisol, prednisone, methylprednisolone, fludrocortisone, prednisolone, dihydrotestosterone, corticosterone, dexamethasone, cortisone, pregnenolone, 11-Deoxycorticosterone, aldosterone, progesterone, 21-deoxycortisol.

Factors known to significantly affect the results:

- Poor spot quality will affect the results obtained. Samples that are not uniformly saturated with blood, poorly collected, improperly dried or contaminated samples reported as insufficient for analysis.

(Last updated January 2020)