

## Disorders of sexual development in neonates: investigation

### Protocol Responsibilities and Authorisation

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### Protocol Review History

Version	Updated by	Date Updated	Description of Changes
2	David Bouchier	22/11/16	None

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### 1. Overview

#### 1.1 Purpose

To investigate disorders of sexual development

- (i) Age <36 hrs - U & E, glucose, 17-oH progesterone, karyotype  
- LH, FSH, Antimüllerian hormone, Testosterone
- (ii) Age >36 hrs but <6weeks  
- U & E. glucose, karyotype, LH, FSH, Antimüllerian hormone

HCG Stimulation Test (1000iu  $\equiv$  38.5 mcg)

Day 1 - Take blood for 17-OH progesterone, testosterone, androstenedione, DHEAS, 11-deoxycortisol  
- Administer 1000 iu HCG IMI

Day 2 - Administer 1000 iu HCG IMI

Day 3 - Administer 1000 iu HCG IMI

Day 4 - Take blood for 17-OH progesterone, testosterone, androstenedione, DHEAS, 11-deoxycortisol

- (iii) Age 6-12 weeks - U & E, glucose, karyotype, LH, FSH, 17-OH progesterone, AMH, Testosterone, Androstenedione, DHEAS, 11-deoxycortisol
- (iv) Assess genital structure - pelvic U/S or MRI.

Sample	Tube (green top)	Min vol (mL)
U & E/glucose (1)	heparin	0.3 mL )
LH/FSH	heparin	0.2 mL )
Testosterone (3)	heparin	0.2 mL ) min 1.2mL (preferably 2 mL)
11-deoxycortisol	heparin	0.2 mL )
Antimüllerian hormone	heparin	0.3 mL )
17-OH progesterone (2)	plain	0.3 mL )
DHEAS	plain	0.2 mL ) min 0.7 mL (preferably 1 mL)
Androstenedione	plain	0.2 mL )
Karyotype (4)	heparin	1 mL in standard adult 4 mL tube

Total volume = 4mL  
(Number = priority of tests)

*If unable to achieve blood sample volumes, do tests in the numerical order shown above.*