

British Society for Paediatric Endocrinology and Diabetes.

Antenatal assessment and treatment of women with a previously affected child with Congenital Adrenal Hyperplasia (CYP21) and the subsequent monitoring of outcome parameters post delivery.

Proforma 1: Laboratory Data (Molecular biology)

Name of Index case
Sex of Index Case Male/Female
Reference Number
Referral Centre
Consultant (Paed Endo)

Laboratory Used

London/Manchester

Antenatal assessment of the family.

INDEX CASE:	Mutation found in CYP21	Yes/No
	Details if Yes
MOTHER:	Mutation found in CYP21	Yes/No
	Details if Yes
FATHER:	Mutation found in CYP21	Yes/No
	Details if Yes

Genetic Studies of CVS

Gestation:weeks
Karyotype:
Mutation found in cyp21: Yes/No
Details if Yes:

Confirmatory Studies of Fetus

Cord blood Yes/No
Karyotype
Mutation found in CYP21 Yes/No
Details if Yes

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Proforma 2: Genetic data

Name of Index case
Sex of Index Case Male/Female
Reference Number
Referral Centre
Consultant (genet)

Antenatal assessment of the family.

INDEX CASE:	Genital Ambiguity	Yes/No
	Salt wasting	Yes/No
	Mutation found in CYP21	Yes/No
	Details if Yes
MOTHER:	Mutation found in CYP21	Yes/No
	Details if Yes
FATHER:	Mutation found in CYP21	Yes/No
	Details if Yes
FH of CAH	Sibling	Yes/No
	Other (specify)
Antenatal diagnosis offered		Yes/No
Antenatal diagnosis accepted		Yes/No
Antenatal treatment offered		Yes/No
Antenatal treatment accepted		Yes/No

Genetic Studies of CVS

Gestation:weeks
Karyotype:
Mutation found in CYP21: Yes/No
Details if Yes:
Treatment stopped Yes No

Confirmatory Studies of Fetus

Cord blood taken Yes/No
Karyotype
Mutation found in CYP21 Yes/No

Dose red										
Psych Q										

Key

Pr-uria: Urine check for proteinuria

Pl oestr: Maternal plasma unconjugated oestriol (JH-see protocol) {fetal suppression}

Ur t oestr: Urinary total oestriol [24 hrs] (JH- see protocol)

Ur Cmet: Urinary cortisol metabolites [24hrs] (JH- see protocol)

Glycosur: Urine check for glycosuria

** OGTT at 28weeks *only*, unless clinically indicated

Dex dose: dose of dexamethasone (mg/kg) [write "stop" if discontinued & state reason]

Dose redn: dose of dexamethasone reduced (Yes/No) [State reason below

Psych Q: Psychological Questionnaire performed (Yes/No)

Dexamethasone

Date started

Date stopped

Reason stopped

Dose reduced Yes/No

Reason reduced

Fetal Monitoring

CVS atweeks

Karyotype

CYP21 mutation Yes/No (see *Proforma 1. Laboratory data*)

Deaxamethasone Continued/Discontinued

Data	Booking	20 weeks	28 weeks	34 weeks
Growth				
Adrenal Suppression*				

* Adrenal suppression: See above, *Maternal plasma unconjugated oestriol*

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Proforma 4: Paediatric Endocrine data

Name of Index case
Sex of Index Case Male/Female
Reference Number
Referral Centre
Consultant (Paed Endo)

Index Case

Age of diagnosis
Salt wasting Yes/No
Genital Ambiguity Yes/No
Prader Stage (1-5)
Photography Yes/No

Current Pregnancy

CVS Yes/No
Karyotype
CYP21 Mutation Yes/No

Dexamethasone Yes/No
Date started
Date stopped
Reason stopped

Fetal Monitoring

Normal growth Yes/No
Details if no
Adequate suppression Yes/No
Details if no

Post-partum

Normal delivery Yes/No
Details if no
Gestationweeks
Birth weightKg

SGA Yes/No
Placental weightgms

Affected female Yes/No
Affected male Yes/No
Genital ambiguity Yes/No
Prader stage (1-5)
Photography Yes/No

Comparison of genital anomaly with index case

Similar Yes/No
More severe Yes/No
Less severe Yes/No

Other congenital anomaly found Yes/No
Details if Yes

Post partum serology

Cord blood taken Yes/No
Genetic studies Yes/No
If Yes, result
Discrepancy with CVS Yes/No
Serum sample saved Yes/No

Biochemistry

Pre-feed glucose checked from birth Yes/No
Evidence of hypoglycaemia (<2.6mmol/l) Yes/No
Daily U & E checked Yes/No
Evidence of Salt wasting Yes/No

17 OH Progesterone checked (day 3) Yes/No
Result

Urine Steroid Profile (JH) sent (day 3) Yes/No
Result

Plasma Renin Activity checked (day 3) Yes/No
Result

Short Synacthen performed Yes/No
Basal valuenmol/l
Peak responsenmol/l

Affected infants only

Hydrocortisone start date
9 alpha Fludrocortisone start date
Genital surgery required Yes/No
Details if yes

Long term follow up (ALL infants)

Growth (growth charted separately)

Neurodevelopment (D Skuse) Yes/No
Date performed
Age of child,yrs
Result

Psychosexual development (M Hines) Yes/No
Date performed
Age of Child,yrs
Result

Age 5 -6 yrs

MRI scan Yes/No
Date performed
Age of child,yrs
Result

Oral GTT Yes/No
Date performed
Age of child,yrs
Result

24 hr ambulatory BP Yes/No
Date performed
Age of child,yrs
Result