406

**NEONATE WITH MIXED GONADAL DYSGENESIS:
CHALLENGES IN SEX ASSIGNMENT**Shi Yun Chia, Rashida Farhad Vasanwala. *Singapore*

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Background 45,X/46,XY mixed gonadal dysgenesis (MGD) is a disorder of sex development characterized by a broad phenotypic spectrum. Patients may have unilateral, bilateral or no testis, streak gonads and/or persistent Mullerian structures. It poses a great clinical challenge due to known effects on growth, hormonal balance and gonadal development.

Objectives We present a case of 45,X/46,XY mixed gonadal dysgenesis (MGD) and the challenges of gender assignment discussion with the patient's family

Methods Clinical Case Report

A term new-born infant, delivered at home, presented to the neonatal unit with ambiguous genitalia. Examination revealed a 1.5 cm midline phallic structure, with labial-scrotal folds with rugosity but no urethral opening (figure 1), two external openings at introitus (urethral and vaginal) (figures 2 and 3), and bilateral palpable inguinal gonads, right larger than left.

Results Ultrasound of the pelvis showed a uterus, cervix and vagina with possible right intra-abdominal testis. The neonate passed a synacthen test and had normal 17-Alpha-Hydroxyprogesterone. Gonadotropins (Follicle stimulating hormone and Luteinizing hormone), testosterone and Anti-mullerian hormone were normal but there was no detectable oestradiol. Cytogenetic investigation included FISH and karyotype which showed 45,X[22]/46,X,idel(Y)(q11)[8].ish idel(Y)(p11.3)(SRYx2), of which 25% of cells were Y-containing, while 75% were X-containing. Exploratory laparoscopy with biopsy of bilateral gonads showed right gonadal tissue (figure. 4) and left hemi-uterus and fallopian tube with no ovary (figure. 5). Histology confirmed right testicular tissue and left sided structure resembling fallopian tube showed no ovarian stroma, primordial follicles, nor seminiferous tubules. Findings and options regarding sex of rearing, surgical and medical treatment, were discussed in the family conference. The structure of discussion is appended in table 1.

Conclusions The management of MGD is multi-disciplinary. Gender assignment is based on the consideration of several

factors, including external and internal genital findings, the role of surgical procedures required, future prospects of hormone replacement, fertility, urinary & sexual function and risk of gonadal malignancy. In addition, social and psychological support is important as the family makes the decision on gender assignment.

407

**STUDY ON RETINOPATHY OF PREMATURE –
INCIDENCE, RISK FACTORS & OUTCOME**Ankita Halder, Asha Mukherjee, PK Saraf. *India*

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Background Retinopathy of prematurity (ROP) is a potentially vision threatening disease affecting preterm babies. Progress in neonatal intensive care in recent years has led to an increased survival of preterm & sick babies and subsequently, to an increasing incidence of ROP.

Objectives To analyze the incidence, risk factors and outcome of ROP.

Methods

STUDY POPULATION 50 Babies ≤ 32 weeks gestational age and 50 preterm babies > 32 weeks gestational age.

INCLUSION CRITERIA Babies with birth weight ≤ 1500 g. Babies born at ≤ 32 weeks of gestation. Selected preterm babies with a birth weight between 1500 grams and 2000 grams or gestational age of more than 32 weeks with additional risk factors (eg. oxygen therapy, sepsis, apnea, birth asphyxia, RDS, NEC, use of surfactant, exchange transfusion, IVH, PRBC transfusion).

Group1: Babies with gestation ≤ 32 weeks and/or babies with birth weight ≤ 1500 g.

Group2: Selected preterm babies with a birth weight between 1500 grams and 2000 grams or gestational age of more than 32 weeks with additional risk factors as mentioned above

EXCLUSION CRITERIA Outborn babies treated in our NICU
STUDY PERIOD: 2017- 2018 When to screen: First screening examination should be carried out at 31 weeks of gestation or 4 weeks of age, whichever is later.

Abstract 406 Table 1 Structure of discussion with patient's family

Sex of Rearing	Surgical Procedures Needed	Future Hormone Therapy	Fertility	Sexual Function	Urinary Function	Risk of Cancer in Testes
Female	vClitoral reduction Removal of testes	Estrogen during puberty	vInfertile Can conceive with egg donation but with risk as uterus is abnormal	Intercourse possible	Intact	Nil
Male	vConstruction of penis, scrotal sac and urethra Orchiopexy Removal of uterus and fallopian tubes	May not need testosterone	vSub-fertile Sperm banking may be possible	Penile function affected (erection during intercourse)	May need to sit for urination	Approximately 10%
No assignment currently; decision later	vTo decide at 21 years of age vReconstruction may be more difficult at later age. Genitalia may resemble male appearance as testosterone is active up to 1 year.	Depends on chosen sex	vSub-fertile as male Infertile as female	Depends on chosen sex	Sit for urination currently Requires reconstruction later	Approximately 10%