NEONATE WITH MIXED GONADAL DYSGENESIS: CHALLENGES IN SEX ASSIGNMENT
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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. Cyto genetic investigation included FISH and karyotype which showed 45,X[22]/46, X,dic(Y)[q11][8]; ish Y[pl1.3] (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, primitive 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.

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

<table>
<thead>
<tr>
<th>Sex of Rearing</th>
<th>Surgical Procedures Needed</th>
<th>Future Hormone Therapy</th>
<th>Fertility</th>
<th>Sexual Function</th>
<th>Urinary Function</th>
<th>Risk of Cancer in Tests</th>
</tr>
</thead>
<tbody>
<tr>
<td>Female</td>
<td>vClitoral reduction</td>
<td>Estrogen during puberty</td>
<td>vInfertile</td>
<td>Intercourse possible</td>
<td>Intact</td>
<td>Nil</td>
</tr>
<tr>
<td></td>
<td>Removal of testes</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Male</td>
<td>vConstruction of penis, scrotal sac and urethra</td>
<td>May not need testosterone</td>
<td>vSub-fertile</td>
<td>Penile function affected (erection during intercourse)</td>
<td>May need to sit for urination</td>
<td>Approximately 10%</td>
</tr>
<tr>
<td></td>
<td>Orchidopexy Removal of uterus and fallopian tubes</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>No assignment currently; decision later</td>
<td>vTo decide at 21 years of age vReconstruction may be more difficult at later age.</td>
<td>Genitilia may resemble male appearance as testosterone is active up to 1 year.</td>
<td>Depends on chosen sex</td>
<td>vSub-fertile as male</td>
<td>Depends on chosen sex</td>
<td>Requires reconstruction later</td>
</tr>
</tbody>
</table>

STUDY ON RETINOPATHY OF PREMATURITY – INCIDENCE, RISK FACTORS & OUTCOME
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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 (e.g. 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.
All the babies found to have ROP were regularly followed up in both our high risk clinic and in Ophthalmology outpatient department for one year.

**Results** Incidence of ROP was 23% in our study. Incidence was 38% in the 1st group & 8% in the 2nd group.

Most common findings were stage III ROP (39%) and zone II ROP (70%). 3 (13%) had APROP (Aggressive posterior ROP) and 2 (8.7%) had retinal detachment.

The incidence of ROP increased as the birth weight and period of gestation decreased- no ROP was found in gestation ≥35 weeks and birth weight >1.828 kg.

In our study, oxygen administration through mechanical ventilator or CPAR, sepsis, therapy with surfactant, apnea, PRBC transfusion, NEC and birth asphyxia were found to be significant risk factors. The risk of ROP is more in RDS, IVH and only head box oxygenation but the risk is not significant. We have not found any ROP in the babies who have undergone exchange transfusion.

13 (57%) babies had spontaneous regression of ROP and rest 10 (43%) required some intervention. To summarize the intervention (n=10), 5 (50%) responded to LASER only, 3 (30%) required intravitreal injection with Bevacizumab (anti-VEGF) following LASER, 1 (10%) required TPPV (Trans Pars Plana Vitrectomy) in left eye for ROP stage IVb along with LASER and 1 (10%) baby required all the three- LASER, intravitreal Bevacizumab and left sided TPPV.

**Conclusions** Thus the need for a routine screening program for the detection of ROP in preterm and sick neonates with risk factors is very essential in our clinical settings. Prevention of blindness from ROP can be very effective through early detection and urgent treatment. This needs awareness among neonatologists and pediatricians for referral to the ophthalmologists at appropriate age of the baby.