demographic data, clinical and laboratory data along with the prescribed therapy.

Results From the 1020 studied cases in 260 there was a diagnosis of indirect hyperbilirubinemia, associated pathology consisted of urinary tract infection in 15 cases, piodermatitis in 12, otitis media in 7, acute diarrhoea in 14 cases and severe dehydration in 9 cases.

Only one case complicated with kernicterus. From the 260 cases only 60 had phototherapy in the treatment schedule.

Conclusion we have shown that one quarter of the jaundice at newborn is due to indirect hyperbilirubinemia and that phototherapy is still a useful treatment.

PO-0675 PREVALENCE OF MUTATION C.11864G >A (P. TRP3955X) IN THE USH2A GENE IN PATIENTS WITH USHER II SYNDROME FROM VOLGA-URAL REGION OF RUSSIA

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Usher Syndrome (US) is an autosomal recessive condition characterised by a combination of congenital hearing impairment and retinitis pigmentosa. To date, ten genes have been associated with US, representing up to 90% of cases. Three types of US are known and differ by onset of the symptoms, severity and progressiveness of deafness and additional vestibular dysfunction. Patients with type II US have congenital bilateral sensor neural hearing loss that is mild to moderate in the low frequencies and severe to profound in the higher frequencies, intact vestibular responses, and bilateral retinitis pigmentosa.

40 unrelated Usher II type families (60 patients) from Volga-Ural Region of Russia were studied using genotyping micro array (Usher, Asper-Biotech) for screening 614 mutations in genes *CDH23*, *MYO7A*, *PCDH15*, *USH1C*, *USH1G*, *USH2A*, *GPR98*, *CLRN1*, *DFNB31* and automatic sequencing of Usher's genes. Diagnosis was based on pedigree data, ophthalmologic, audio logical and vestibular examination.

We revealed homozygous and heterozygous genotypes for the c.11864G >A (p. Trp3955X) mutation (*USH2A*) in six unrelated families among Russian, Tatar and Chuvash patients with Usher II syndrome. We found four pathogenic mutations in coding region of 8 patients (p. Glu4458fs, p. Trp3955X, p. Glu4078fs, and p. Gly1392X), confirming their clinical diagnosis. The most frequent *USH2A* gene mutation was c.11864G >A (9/80 alleles; 11,25%). Mutation c.11864G >A in heterozygous state was also found in one Russian subject out of 1066 examined individuals from 16 various populations of Eurasia: Bashkirs, Tatars, Chuvashes, Udmurts, Komi-Permyaks, and Mordvins, Russians, Belarusians, Ukrainians, Veps, and Karelians, Abkhazians, Kazakhs, Uzbeks, Yakuts, Altaians. Study was supported by grants (No12–04–00342_a, No12–04–98520_r_vostok_a, 14–04–97002_r_povolgie_a, 14–04–97007_r_povolgie_a,14–04–01741_A).

PO-0676 PREDICTIVE VALUE OF POSTNATAL NASOGASTRIC TUBE AND CHEST/ABDOMINAL XRAY FOR ANTENATAL POLYHYDRAMNIOS AND/OR SMALL STOMACH IN THE DIAGNOSIS OF OESOPHAGEAL ATRESIA-TRACHEO-OESOPHAGEAL FISTULA

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Introduction Oesophageal atresia (OA) ± trachea-oesophageal fistula (TOF) is a rare congenital anomaly which occurs in ~1/3500 total births.

Prenatal diagnosis of OA-TOF is desirable as it may improve patient outcome by optimising the pre- and postnatal management. Therefore, a high prenantal detection rate is desirable. Prenatal ultrasound is one of the modalities used in improving the diagnostic rate. Indirect or non-specific signs of suspected OA-TOF include polyhdramnios (10–20% of which are associated with fetal abnormalities, one of it being OA-TOF), absent/small stomach bubble, a blind ending upper oesophageal pouch and observation of fetal swallowing.

In our institution, all foetuses with Isolated polyhydramnios ± small/absent stomach have postnatal NGT insertion followed by xray if follow up scans remained the same.

Objectives To evaluate the clinical relevance of postnatal NGT insertion followed by xray for prenatal finding of isolated polyhydramnios ± absent/small stomach bubble in a cohort of newborn babies to assess for the diagnosis of OA-TOF.

Methods

- Retrospective cohort review January2011-March2014 of all foetuses/neonates with suspected EA-TOF on prenatal ultrasound (isolated polyhydramnios [deepest pool >8 cm, Amniotic Fluid Index >95th percentile for gestational age and/or small/absent stomach bubble] or perinatally identified to have >1500 mls amniotic fluid and therefore needing postnatal NGT insertion+ Xrays pre-feed were identified.
- Course and tip of NGT, stomach-bubble, vertebral anomalies or any other abnormalities detectable on Xray.
- Time taken from birth to when the postnatal Xrays were taken was calculated (time to first feed).
- Babies were also examined for clinical signs of OA-TOF or other abnormalities.
- Clinical data on all OA-TOF cases that were diagnosed postnatally over the same 3 yr period.
- Exclusion – above two features with other any other abnormalities on antenatal scans.

Results

- Total deliveries over period= 17,200.
- Total no. fulfilling criteria of isolated polyhydramnios ± small/absent stomach only = 48.
- No. that resolved antenatally and therefore did not have NGT +Xray assessment postnatally = 12.
- Total No. who had NGT+Xray assessment = 36.
- No. without OA-TOF = 35.
- No. diagnosed with OA-TOF = 1.
- 7 cases of OA/TEF (1 had above 2 features); of which 3 did not have polyhydramnios ± absent/small stomach; 3 had multiple abnormalities including 2 with polyhydramnios.
- Above two antenatal features had a Sensitivity =25%; Specificity =99.8%; positive predictive value=2.78% for diagnosis of OA-TOF.
- Mean time for NGT+Xray (away from mothers and before their first) = 156 mins (range 81–332).