Abstracts

648 THE COMPLEX PROBLEMS OF CHILDREN AND FAMILIES WITH A CHILD WITH DISORDERS OF SEXUAL DEVELOPMENT
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Background The diagnose of DSD is a challenge for medical staff, family and society.

Material and method We study 15 patients with DSD, between 2005–2011. The study protocol included anamnesis, clinical examination: auxiological dates, degree of puberty (Tanner), genitalia conformation (Prader stages) and its consistency with social sex. Laboratory datas: karyotype, gonadotrophins, testosterone, DHT, inhibin B, DHEA and DHEAS, 17O progesterone; SRY gene (in selected cases) was performed. In all the cases we perform psychological exams of the child and family.

Results We have eight, 46 XX, and three 46 XY subjects. According to age we have 4 groups: newborns (2), children between 1–3 years(4), between 3–6 years (6) and more than 10 years(2). The diagnosis was: CAH 8 cases, PAIS 1 case, Smith-Lemli-Opitz-Syndrome 1 case, Leyding cell hypoplasia 1 case. In neonatal period the correct assessment of social sex reveal minimal psychological familial problems. Adolescent CAH have general psychological distress higher rates of substance abuse, somatization, and suicidal behaviors. PAIS child want to change “her” sex because poorer social and interpersonal relationship functioning. The family doesn’t agree because they live in a community with specific rules.

Conclusions
1. In DSD cases the assessment of sex must be done in neonatal period after careful evaluations of the child.
2. The psychological implications are more deep and affect the child, family and the society.
3. The right of the child must be protected by specific low of the state.

649 A CASE OF PERMANENT NEONATAL DIABETES MELLITUS
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Background and Aims Neonatal Diabetes Mellitus(NDM) is a rare(1/400,000 newborns) but potentially devastating condition. It has been defined as insulin-sensitive hyperglycemia that is diagnosed within the first six months of life and can be either transient(TNDM) or permanent(PNDM). PNDM has been linked to mutations in several different genes. TNDM is associated with defects in an imprinted region of the paternally derived chromosome 6. We describe a baby boy four months old diagnosed with PNDM.

Methods The patient was admitted to hospital with diabetic ketoacidosis(blood sugar>700mg/dl (=38.8mmol/l), pCO2,22mmHg, pO2,107mmHg, HCO3,12.1mmol/l). He had a preceding fortnight history of polyuria, polydipsia, lethargy and vomiting the last few days before admission. Clinically he was lethargic and dehydrated, with sunken fontanelle and eyes, reduced skin turgor, dry mucous membranes and had tachypnoea, ketotic breath. He was treated with intravenous fluids and insulin. Progressively he recovered and started feeding orally. He was discharged on daily insulin injections subcutaneously.

Results On admission glyced hemoglobin(HbA1c) was 5.8% (4.3–6.1%), anti-GAD autoantibodies 1.3(ratio=1.1 positive), IA2(tyrosine phosphatase antibodies) 1.2(< 8iu/ml), IAA(anti-insulin antibodies) 1.50(< 1.10 ratio), ICA(anti-islet antibodies) 0.5(< 1.00 ratio), EMA(anti-endomysial antibodies) negative. Past medical and family history were unremarkable. Genetic testing for PNDM failed to detect any mutations in the KCNJ11, ABCC8 and INS genes, as well as the testing for abnormalities in the chromosome 6q for TNDM.

Conclusions Genetic testing for NDM can identify PNDM in newborns helping the physician to select the most appropriate therapy. However, 40% of cases are currently without a molecular genetic diagnosis.

650 UMBILICAL CORD AND FIFTH DAY SERUM VASPIN CONCENTRATIONS IN SMALL, APPROPRIATE AND LARGE FOR GESTATIONAL AGE NEONATES
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Background and Aim Vaspin is a visceral adipose tissue derived serin protease inhibitor which has an insulin sensitizing effect. It is correlated with insulin resistance and glucose metabolism and it improves glucose tolerance. Our aim was to determine and compare serum vaspin and insulin concentrations in small-for-gestational age [SGA], appropriate-for-gestational age [AGA] and large-for-gestational age [LGA] infants at birth and fifth day of life.

Methods Eighty-two neonates were divided into three groups, as SGA [n=22], AGA [n=30] and LGA [n=30]. Mothers age, gestational week, mode of delivery, maternal diseases like diabetes, preeclampsia and eclampsia were recorded. Blood for vaspin, insulin and glucose was collected from cord at birth and peripherec vein on the fifth day of life.

Results At birth, there were no statistically significant difference in serum insulin concentrations between the three groups whereas cord serum vaspin concentrations were significantly higher in SGA group [p<0.05 & p<0.005]. Serum glucose and vaspin levels on postnatal 5th day of life had no significant difference between three groups [p>0.05]. Circulating vaspin concentrations were not associated with sex of the infant and delivery route.

Conclusion Cord vaspin levels are significantly higher in SGA neonates than AGA or LGA neonates. The fetal programming hypothesis proposes that many adulthood diseases originate through adaptation which the fetus makes when it is undernourished. High cord vaspin levels in SGA infants may be one of the adaptation for increased risk for adult metabolic diseases.

651 EVALUATION OF THYROID FUNCTIONS IN PRETERM NEWBORNS LESS THAN 34 WEEKS OF GESTATION
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Objective To evaluate the thyroid functions of preterm infants.

Methods Data about thyroid functions (FT4, TSH) were collected retrospectively from 428 preterm infants less than 34 weeks of gestation, who were born between 2006 and 2009.

Results The mean gestational age of the study group was 30.5±2.4 (23.9–33.9) weeks, the mean birth weight was 1339±496 (496–3190)
and during childhood period (4.5/−2 years) of age. Placental weight (BW), and head circumference (HC) were obtained at birth using conditional analysis. Placental weight, infant length (BL), results of our study with those stated in the references. Normal sensitivity therapy by referral Endocrine Diseases Clinic and auditory brainstem responses test (ABR) was performed for all subjects. One of the associated abnormalities in these patients is the sensorineural hearing defect, which has a prevalence of about 1%. The study group was divided into three subgroups according to gestational weeks (g. The study group was divided into three subgroups according to gestational weeks (< 28 wk, n=79; 28–31 6/7 wk, n=204; 32–33 6/7 wk, n=145). Twenty five percent of the infants were small for gestational age (SGA). Mean age at first thyroid function evaluation was 18.3±12.5 days. Mean FT4 levels were 12.0±3.1, 14.1±3.3 and 17.7±3.9 pmol/L in three subgroups, respectively and significantly lower in infants < 28 weeks. In all subgroups SGA infants had lower FT4 levels, but it was significantly lower in only 28–31 6/7 and 32–33 6/7 weeks but not in < 28 weeks subgroup. Overall, the prevalence of hypothyroxinemia and hypothyroidism were 25% and 0.8%, respectively in the first evaluation. 17.6% of infants < 28 weeks had hypothyroxinemia (n=153) and all of them were treated. In the total group levothyroxine treatment was given to 51 (11.9%) infants. Mean treatment period was 1.6±1.2 years.

Conclusion Free T4 levels were lower in the early gestational age subgroups. SGA infants had lower FT4 levels.

652 PREVALENCE OF SENSORINEURAL HEARING LOSS IN PATIENTS WITH CONGENITAL HYPOTHYROIDISM doi:10.1136/archdischild-2012-302724.0652

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Background and Aims Congenital hypothyroidism is mainly diagnosed through neonatal screening program. Normal physical and mental development can be maintained with pertinent replacement therapy. One of the associated abnormalities in these patients is the sensorineural hearing defect, which has a prevalence of about 20% according to relevant references. The purpose of this study was to obtain the prevalence of sensorineural hearing loss in children with congenital hypothyroidism identified in the screening program in Qazvin, Iran.

Methods All patients afflicted with congenital hypothyroidism identified in the screening program (in Qazvin, Iran) were enrolled in this study. They were both under observed and hormonal replacement therapy by referral Endocrine Diseases Clinic and auditory brainstem responses test (ABR) was performed for all subjects.

Results Of 169 patients with congenital hypothyroidism, 42.3% were female. The prevalence of sensorineural hearing loss was 5.3% (6 male, 2 female). Statistical analysis did not reveal any significant difference between the prevalence of sensorineural hearing loss with other variables of the study.

Conclusions A remarkable difference was observed between the results of our study with those stated in the references. Normal sensorineural hearing can be maintained with pertinent replacement therapy.

653 PLACENTAL WEIGHT: RELATION TO MATERNAL WEIGHT AND GROWTH PARAMETERS AT BIRTH AND DURING CHILDHOOD doi:10.1136/archdischild-2012-302724.0653

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Background Human growth is a continuous process. Studies defining placental effect on growth focus on discrete time points (e.g., birth), overlooking the conditional nature of the process.

Material and Methods Two hundred mothers who gave birth at term after an uncomplicated singleton pregnancy were studied using conditional analysis. Placental weight, infant length (BL), weight (BW), and head circumference (HC) were obtained at birth and during childhood period (4.5/−2 years) of age. Placental weight was correlated with growth parameters of the child at birth and during childhood.

Results At birth, placental weight was correlated significantly with maternal weight (r=0.21, p=0.031), infant BW (r=0.71, r < 0.001), BMI SDS (r=0.589, p<0.001), LS SDS (0.567, p<0.001), and HC (r=0.699, p<0.001). During childhood, placental weight was correlated with BMI SDS (r=0.296, p=0.002), Ht SDS = (r=0.254, p=0.009). Length SDS at birth was correlated significantly with Ht SDS during childhood (r=0.445, p<0.001).

Conclusion Placental weight is a good pointer of birth size (weight, length and HC) and may help forecast childhood growth.

654 THE DIFFERENTIAL-DIAGNOSTIC FEATURES OF THELARCHE IN GIRLS doi:10.1136/archdischild-2012-302724.0654

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Aim To determine the most significant criteria in the diagnosis of true precocious puberty and premature thelarche.

Methods 68 girls were analyzed in the endocrinological department in Minsk over 2003–2011 yrs. Group 1 (G1) - girls with isolated thelarche (IT) (58 (85.3%); group 2 (G2) - with true precocious puberty (TPP) (10 (14.7%). Ultrasound (u/s) organs of the small pelvis, bone age, the levels of hormones (follicle-stimulating hormone (FSH), luteinizing hormone (LH), estradiol (E2); gonadotropin-releasing hormone analogue (GRH) stimulating test were conducted to all patients. Results were processed using the Statistica 6.1.

Results Breast development in G1: stages on Tanner 2 (84.3%), 3 (15.7%); G2: Tanner 2 (80%), 3 (20%). The onset of thelarche G1 1.20±0.3 yrs, G2 5.5±0.77 (p=0.2). Bone age (Boa)/biological age (Bia) G1 0.63±0.08 (1 yrs), G2 3.3±0.01. Uterus length G1 28±0.83 (35 mm), G2 35.4±2.9 (p=0.1). There was an excess of prepubertal ovaries norm (>0.2ml) G1 65%, G2 48% with the presence of follicles G1 20%, G2 100%. Basal FSH levels G1 4.73±0.52 (1.8–10.5 IU/L), G2 4.25±0.87 (p=0.3); LH G1 0.76±0.13 (1–10 IU/L), G2 2±0.8 (p=0.15); E2 G1 0.12±0.02 (< 0.5 ng/ml), G2 0.14±0.05 (p=0.08). There was a pubertal excess of LH levels in G2 (39.4±20.4 IU/L) by conducting GRH stimulating test.

Conclusions The differential diagnosis between TPP and IT are: advance Boa to BiA, the excess of uterus length by u/s and E2 levels, excess of LH levels by conducting stimulating test with GRH (which is the most important feature).

655 GENDER PECULIARITIES OF THE COURSE OF GRAVES-BASEDOW DISEASE IN CHILDREN doi:10.1136/archdischild-2012-302724.0655

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Aim To determine gender peculiarities of the course of Graves-Basedow disease (GBD) in children depending on the sex and stage of puberty.

Methods We analyzed retrospectively 28 children with GBD in the endocrinological department of University hospital over the 2011 year (boys(B)/girls(G) 3/25, mean±SD age 13.15±2.42 yrs) (ð=0.05). The onset of GBD was mainly in late puberty regardless of gender. There was lower TSH 0.085±0.13 (0.23–3.4mlU/L) and higher free T4 62.13±34.34 (10.23.2pmol/l) levels in B (G 0.12±0.2 and