

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, 17OH 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, Simith-Lemli-Optz-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 law 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 glycated hemoglobin (HbA_{1c}) 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 peripheral 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 [$\chi^2=8,158$ p<0.05]. Serum glucose and vaspin levels on post-natal 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)