OC8

HIGH PREVALENCE OF *WT1* GENE MUTATIONS IN PATIENTS WITH STEROID-RESISTANT NEPHROTIC SYNDROME

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Background and aims Steroid-resistant nephrotic syndrome (SRNS) is the rare disease and holds a special place in structure of nephrotic syndrome in children, it takes up to 15–25% among other forms of the nephrotic syndrome. SRNS is one of the most common causes of end-stage kidney failure. The majority of SRNS cases are caused by genetic mutations leading to structural and functional glomerular injuries. Treatment strategy in children with SRNS is defined by results of molecular genetic testing and morphological diagnosis after kidney biopsy. Research is focused on identification of pathogenic gene variants leading to kidneys hereditary diseases development united by SRNS symptoms complex.

Methods The research included 83 patients (40 girls, 43 boys) aged from 0 up to 16 years with SRNS who undergone molecular genetic testing by next generation sequencing (NGS) with further results validation by Sanger sequencing and bioinformatics analysis by means of the program Alamut.

Results Mutations in *WT1* gene were revealed in 13 cases (10 girls, 3 boys, mean age — 7,5 years, manifestation mean age — 2 years), four patients have congenital nephrotic syndrome. Revealed morphological variants: focal segmental glomerulosclerosis in 10 cases, minimal changes disease — 3 cases. Pathogenic variants of *WT1* gene: 6 missense (*c.1217G>A*, *p.R406K*; *c.1270A>G*, *p.K424E*; *c.1311T>G*, *p.F437L*; *c.1399C>T*, *p.R467W*; *c.1407C>A*, *p.D469E*; *c.1433A>G*, *p.H478R*) and 2 splicing (*c.1447+4C>T*, *c.1447+5G>A*) mutations (NM_024426.5 transcript). We revealed two novel variants (*c.1217G>A*, *p.R406K*; *c.1311T>G*, *p.F437L*) undescribed in HGMD professional database. The most common mutation in *WT1* gene is *c.1447+5G>A*. Mutations in *WT1* gene cause various clinical phenotypes: Denys-Drash syndrome (8%), Frasier syndrome (15%), isolated SRNS (77%). All patients with *WT1* mutations have significantly lower levels of proteinuria against whole patient cohort, nearly all have sustained hypertension and intermittent hematuria.

Conclusion It was established that in 16% of all investigated cases the key factors for the manifestation and progression of SRNS are *WT1* gene mutations. Mutations *c.1399C>T* and *c.1433A>G*, are associated with rapid progression and early end-stage kidney failure in the age under 6 years.

OC9

IMPACT OF COMBINED ANTIRETROVIRAL PROPHYLAXIS ON HEALTH OUTCOMES IN HIV-EXPOSED NEONATES

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Modern approaches to administration of antiretroviral therapy (ART) to prevent HIV transmission from an infected woman to her child during pregnancy, labor, and delivery (in case she does not breastfeed her child) significantly reduce the risk of newborn infection. Combined ART is indicated for children with perinatal HIV exposure at high risk of being infected and includes administration of three oral antiretrovirals: zidovudine, nevirapine, lamivudine.

The aim To assess the health of newborns, who underwent the combined antiretroviral therapy to prevent perinatal HIV transmission.

Materials and methods We conducted a post-hoc analysis of 163 cases of HIV-positive women and their newborns at Irkutsk Perinatal Centre (tertiary medical centre), covering 2017–2018 years. Inclusion criteria were high risk of perinatal HIV transmission (a woman did not take antepartum ART, unknown HIV viral load or viral load near delivery is more than 50 copies/ml), a positive result of rapid (point-of-care) HIV antibody test in the mother during delivery, and epidemiological parameters. All newborns with perinatal HIV exposure were given three drug regimen, were formula-fed, and their blood was tested by means of polymerase chain reaction. The follow-up of newborns included clinical monitoring, assessment of hematologic (anemia, neutropenia) and other adverse effects of ART.

Results Out of 445 newborns with perinatal HIV exposure, 163 (36.6%) neonates underwent combined ART. The most frequent causes of combination ART prescription were: no adherence (34.8%) to or no taking (39%) ART by the mother of the neonate during pregnancy, high HIV viral load (17.2%), and epidemiological indications (parenteral substances use during pregnancy – 9%). Fifty-six (34.3%) cases resulted in premature births. An average HIV viral load near delivery was 2800 [150; 8100] copies/ml. Average body weight of newborns was 2590 [700; 4180] grams. Among adverse clinical effects of ART in 60% of prematurely-born HIV-exposed neonates, symptoms of enteropathy prevailed; 19.7% of newborns developed severe anemia by the 28th day after birth. HIV RNA was detected in 1 (0.6%) newborn of this group. Four (2.4%) newborns with perinatal HIV exposure died.

Conclusions Lack of social and medical support of reproductive women, belonging to risk groups, increases the risk of perinatal HIV transmission. Personalized approach to ART planning helps not only to reduce the risk of mother-to-child transmission, but also avoid toxic side-effects of preparations on neonates with perinatal HIV exposure.

OC10

'I HAVE A DEAD PERSON'S KIDNEY IN ME': THE STRUGGLES OF SELF-IDENTITY AMONG ADOLESCENT KIDNEY RECIPIENTS IN IRELAND

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Objective Chronic Kidney Disease (CKD) is a complex disease that requires lifelong treatment. Transplantation is the goal standard treatment, however; it is not without its challenges. While previous research has demonstrated that transplantation increases survival rates and enhances young people's quality of life, little is known about the impact of kidney

transplantation on adolescents' self-identity development. This presentation will discuss the struggles that adolescent transplant recipients experience as they endeavour to incorporate a new kidney into their embodied self. These findings were identified as a predominant theme from a narrative inquiry of embodiment experiences among young people living with CKD in Ireland.

Methods This study employed a narrative inquiry research design. Using an ethnographic approach data were gathered through multi-media approaches including: visual body maps (i.e. life-size human body images), oral informal conversations and observational field notes. Five adolescents (10–17 years) participated in the study, which took place over a period of 18 months as adolescents experienced living with CKD in both hospital and home settings.

Results A nuanced finding identified from this study was a disrupted sense of self and identity arising from adolescents' encounters with a foreign body part (i.e. kidney). Adolescent participants experienced a tension between their body, self and identity as a consequence of transplant surgery. They struggled to adjust to the presence of a foreign organ from a deceased donor and they were concerned about developing personality characteristics of the donor. As one participant described, the kidney was not viewed as her own but was conceptualized as 'a dead persons kidney'. Another participant described the prospective of acquiring the personal qualities or characteristics of the donor as 'disgusting'. Findings also revealed that adolescents engaged in different individualised ways to cope with the transplanted organ.

Discussion The findings from this study provide an understanding of the struggles adolescents endure as they attempt to incorporate a foreign organ within their bodies and the potential impact on their self-identity. Clinicians need to be aware that substituting kidneys with donor organs is much more than a complex technical procedure; it requires an amalgamation of 'other' and 'self'. From a clinical perspective, a multifaceted and holistic person-centred care approach is needed to equip adolescents with the individual skills required to adapt to a new kidney.

OC11 CURRENT BREAST FEEDING PRACTICES AND FACTORS AFFECTING ITIN AL-BAHA REGION, SAUDI ARABIA(PRE & POST – INTERVENTION)

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Introduction and aim of the work Breast feeding (B.F.) is now considered a basic human right for every newborn infant, as well as of his/her mother. The WHO and UNICEF have emphasized the importance of promoting B.F. as a way to improve the health and nutrition of infants and young children.

The objective of this study was to find out the prevalence of B.F., the social, demographic and medical influencing factors related to the initiation and continuation of B.F., the knowledge and attitude of females towards B.F. in Al-Baha, Saudi Arabia. Also, to determine the weak points and common misbeliefs, which should be considered during the intervention stage.

Subjects and methods The study was conducted in Al-Baha region, Saudi Arabia (twice) in the period between January and November, 2015 (pre-intervention) and 2016 (post-intervention). Pre-intervention study was followed by intervention plan in form of workshops for physicians and mothers, and establishment of breast feeding clinic in KFH and use of educational materials in Al Baha hospitals. In both studies, participants were 714 mothers having 714 infants below 24 months, from both sexes and equally distributed among six age groups. All the sampled mothers completed answering questionnaires.

The results The results showed remarkable improvement in all breastfeeding rates, but still far below international safe rates. Post- intervention versus pre-intervention results showed: significantly less percent of mothers who never breast feed, increase ≥ 4 times in the percent of mothers who had BFD = (30–120) and BFD = (120– 240) and similar percent of mothers who had BFD higher than 240 days. Regarding education of the mothers, there was significant increase in BF rate specially in mothers who cannot read or write and those whose education level is before high school. Also the increase in BF rate was more in working mothers (3times) and in mothers from families with high income than non working ones and those from low income families respectively. The factors most significantly associated with the outcome of breast feeding were the early supplements, misperceptions, mother's education, working mothers, baby refusal, contraceptives, inconvenience and use of teats. The mother's knowledge about BF was very poor in both studies but improved in post-intervention study.

Conclusion The current study emphasizes that the most significant factors affecting the outcome of breast feeding are highly modifiable by health education. This information is crucial for the development of a successful breastfeeding promotion plan in this community.

OC12 ASSOCIATION BETWEEN DEPRESSION AND CARDIOMETABOLIC RISK FACTORS IN OVERWEIGHT AND OBESE TURKISH ADOLESCENTS

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Objective The aim of this study was to determine the prevalence of depression in obese and overweight adolescents and, to evaluate the relationships between depression and cardiometabolic risk factors in obese and overweight adolescents.

Method We performed a retrospective cross-sectional analysis of the data from overweight or obese adolescents 11 to 18 years of age who were evaluated in our clinic from January 2012 to December 2015. Depression was evaluated by Children's Depression Inventory. Hypertension, dyslipidemia, hyperinsulinemia, hyperglycaemia and insulin resistance were defined as cardiometabolic risk factors. For statistical analysis, Chi-square test and logistic regression analysis were used.