HOW TO SELECT THE SWEAT TEST CANDIDATE. 10 YEARS OF EXPERIENCE IN SCREENING FOR CYSTIC FIBROSIS IN CHILDREN

PO-0359

Background Sweat test (ST) remains gold standard in cystic fibrosis (CF) diagnosis. Alarm symptoms are age-related.

Aims Retrospective review of cases subjected to ST.

Methods Patients were selected by paediatricians, neonatologists, surgeons, based on suggestive symptoms, personal (PH) and familial history (FH). Inclusion criteria: for 0–1 month age group, patients with PH of atelectasis, meconium ileus, intussusception; 1–12 months, recurrent wheezing (RWF), failure to thrive (FTT); 1–5 years, previous group symptoms, plus chronic cough/diarhoea; >5 years, 1–5 years symptoms, plus recurrent pancreatitis/sinusitis. For all age, patients with PH of salty taste of sweat (STS), salt wasting syndrome, heat shock (HS), and FH of CF, azoospermia. ST was performed with Nanoduct system. Values >60 mmol/L (equivalent NaCl) were considered normal, between 60–80 equivocal, >80 positive. Normal values patients were eventually retested, those with equivocal at least once, those with positive mandatory twice.

Results Were performed 406 ST (344 patients): at 0 month 11 tests (45,4% peritonitis), at 1–12 months 173 (65,3% RW), at 1–5 years 108 (25,9% FTT), at >5 years 50 (62,3% repeated pneumonia-RP). 4 tests equivocal, 5 false positive, 6 positive (5 infants: 1 RW, 1 HS, 2 atelectasis, 1 STS; 1 male 4 years old RP). Relating to age, only 1 positive from 141 RW infants (p 0,03) and 1 from 15 RP children; 2 positive from 6 atelectasis patients and 25 control children of both sexes, with age range 5 years, previous group symptoms, plus chronic cough/diarhoea; >5 years, 1–5 years symptoms, plus recurrent pancreatitis/sinusitis. For all age, patients with PH of salty taste of sweat (STS), salt wasting syndrome, heat shock (HS), and FH of CF, azoospermia. ST was performed with Nanoduct system. Values >60 mmol/L (equivalent NaCl) were considered normal, between 60–80 equivocal, >80 positive. Normal values patients were eventually retested, those with equivocal at least once, those with positive mandatory twice.

Conclusions Great attention on infants with other symptoms than classical ones: higher statistical significance for STS, HS, atelectasis.

BONE MINERAL DENSITY IN EGYPTIAN CHILDREN WITH FAMILIAL MEDITERRANEAN FEVER

PO-0360

Background Familial Mediterranean fever (FMF) has episodic or subclinical inflammation that may lead to a decrease in bone mineral density (BMD).

Objective To assess BMD in Egyptian children with FMF on genetic basis.

Subjects and methods A cross sectional study included 45 FMF patients and 25 control children of both sexes, with age range between 3–16 years old. The patients were reclassified into 2 groups: Group 1 (A) 23 cases used colchicines for 1 month or less, and Group 1 (B) 22 cases used colchicines for more than 6 months. For both patients and control, MEVF mutations were defined using molecular genetics technique and BMD was measured by DXA at 2 sites: proximal femur and the lumbar spines.

Results Four frequent gene mutation where found in the patient group: E148Q (35,6%), V726A (33,3%), M680I (28,9,0%) and M694V (2,2%). There were also 4 heterozygous gene mutations in 40% of control children. Patients received Colchicines treatment for less than 1 month had highly significant lower values of BMD at femur and lumbar spines than control children (p < 0.007, p < 0.001). Patients received Colchicines treatment for more than 6 months had improved values of BMD at femur compared to control, but there were still significant differences between them at lumbar spine (p > 0.036). There are insignificant effect of type of gene mutation on BMD and the risk of osteopenia among the patients.

Conclusion FMF had significant effect on BMD. However; regular use of colchicines treatment improves this effect mainly at femur.

THE SILVER-RUSSELL SYNDROME: REPORT OF 2 CASES

PO-0362

Background The Silver-Russell syndrome is rare pattern of malformations which associated growth retardation generally starting in antenatal period, a characteristic facial feature and limb asymmetry. We report two new cases illustrating this syndrome.

Observation 1 A female infant was admitted in paediatric department at the age of 4 months. The neonatal period was unremarkable; the infant had no intrauterine growth retardation (birth weight and height ranging between the 25th and 50th percentile). The infant’s head circumference was large contrast with