

Rotavirus is the main cause of AGE accounting for about 35% of all causative agents.

About 90% of all cases of Rotavirus AGE occur in children under 5 years of age and

More than 50% of cases occur in infants.

Vaccination against rotavirus is now the best preventive measure.

### 703 CASE REPORT - A PATIENT WITH BONE FORMATION DEFECTS DURING PRENATAL LIFE DIAGNOSED AS 'SHWACHMAN-DIAMOND SYNDROME'

doi:10.1136/archdischild-2012-302724.0703

<sup>1</sup>O Beser, <sup>2</sup>I Dokurel, <sup>1</sup>T Kutlu, <sup>1</sup>T Erkan, <sup>1</sup>FC Cokugras. *Pediatric Gastroenterology, Hepatology and Nutrition; <sup>2</sup>Dept. of Pediatrics, Istanbul University Cerrahpasa Medical Faculty, Istanbul, Turkey*

**Case Report:** Shwachman-Diamond syndrome (SDS) is an autosomal recessive disorder with clinical features that include pancreatic exocrine insufficiency, hematological dysfunction and skeletal abnormalities during development after birth and varies with age. The patient was born as the first child to healthy, term. Labour was induced at 20 wk gestation because of symmetrical fetal growth retardation. Congenital infections were excluded. In the fetal karyotype analysis, no chromosomal abnormalities were detected. Molecular genetic analysis for achondroplasia and hypochondroplasia was performed and gave negative results. After birth, laboratory studies were normal. At the age of six months the patient's height, weight and head circumference remained below 3 percentile. During the sixth month of life, the child's blood count were evaluated retrospectively and intermittent neutropenia was documented. When she presented with increased fat loss and failure to thrive, fecal elastase level was < 50ug/g, low serum trypsinogen level was found. These results were primarily the signs of severe pancreatic insufficiency. On the CT scanning of pancreas, scattered fatty infiltration was detected. The X-ray imaging of the thorax showed narrow thorax. Although on spinal X-ray metaphyseal changes on the costochondral junctions was not detected; 'Metaphyseal dysostosis' of the femoral head and distal femur the was observed more remarkable than upper limbs. When SBDS gene which is a causative gene for Scwachman-Diamond Syndrome was analyzed by DNA sequence analysis.

**Discussion** In contrast to the previous observations, results suggest that the characteristic skeletal changes present in some patients in the intrauterine period of life. Our observation underlines the necessity to consider SDS as the other common reasons of prenatally diagnosed bone disorders like achondroplasia and hypochondroplasia.

### 704 MALROTATION - NOT A LAUGHING MATTER

doi:10.1136/archdischild-2012-302724.0704

E Robertson, M Thorpe. *Paediatrics and Child Health, Royal Cornwall Hospital, Truro, UK*

**Background** Early diagnosis and management of intestinal malrotation can potentially prevent the devastating complications associated with midgut volvulus. Atypical presentation of intestinal malrotation in older children frequently leads to a significant delay in diagnosis.

**Methods** A 6-year-old boy presented with a five year history of hiccups, then vomiting after laughing. Sadly, he had resorted to avoiding laughter. Barium studies demonstrated intestinal malrotation, therefore a Ladd's procedure was performed and he made a good recovery. The case prompted a literature search for aetiology, diagnosis and management of malrotation in children outside of the neonatal period.

**Results** We present a review of this literature, including findings that diagnosis of intestinal malrotation in the older child is

frequently delayed. One study demonstrated a mean delay to diagnosis of 1.7 years. Vague abdominal symptoms including chronic abdominal pain, intermittent non-bilious vomiting, diarrhoea and failure to thrive have been associated with malrotation, but hiccups and laughter have not previously been described. Recent review of diagnostic imaging recommended ultrasound as the investigation of choice, and may advocate its use in the work up of children presenting with non-specific abdominal symptoms. Although debated in the literature, a Ladd's procedure is recommended for all children, even if asymptomatic.

**Conclusion** Children with intestinal malrotation present us with a diagnostic challenge, but a low threshold of suspicion in patients presenting with vague abdominal symptoms, including intractable hiccups or vomiting, may ensure these children receive the timely surgical intervention.

### 705 GASTROINTESTINAL HEMORRHAGE IN A 21-MONTH-OLD GIRL IN THE COURSE OF LYMPHODULAR HYPERPLASIA. CASE REPORT

doi:10.1136/archdischild-2012-302724.0705

A Mroczkowska-Juchkiewicz, A Pawlowska-Kamieniak, D Golycka, K Kominek, E Pac-Kozuchowska. *Medical University, Lublin, Poland*

Gastrointestinal hemorrhage is an emergency condition requiring immediate diagnostics. Lymphonodular hyperplasia, as a cause of bleeding from the gastrointestinal tract in children, is a rarely observed pathology of unknown etiology. The case of a 21-month-old girl is presented with massive bleeding from the lower gastrointestinal tract in the course of lymphonodular hyperplasia. Endoscopy of the upper and lower gastrointestinal tract was performed as an emergency procedure. Gastroscopic examination did not indicate any deviations from the normal. Colonoscopy showed no organic pathology of the mucosa in the terminal ileum, and the presence of blood in the small intestine was not observed. In the entire large intestine the presence of hemolyzed blood was noted and a massive lymphonodular hyperplasia on the mucosa. Histopathologic tests of the specimens taken from the large intestine confirmed normal pit patterns, mucosal edema, presence of lymph follicles, and numerous apoptic bodies in the lamina propria beneath the epithelium and in the surface epithelium.

### 706 INTESTINAL EPITHELIAL DYSPLASIA - A CASE REPORT OF A Milder PHENOTYPE

doi:10.1136/archdischild-2012-302724.0706

<sup>1</sup>G Lesanu, <sup>1</sup>I Tincu, <sup>2</sup>M Gherghiceanu, <sup>1</sup>C Becheanu, <sup>3</sup>M Dumbrava, <sup>1</sup>D Pacurar, <sup>1</sup>V Danila. *'Grigore Alexandrescu' Emergency Children's Hospital; <sup>2</sup>'Victor Babes' National Institute; <sup>3</sup>'Fundeni' Clinical Institute, Bucharest, Romania*

The intractable diarrhea of infancy remains a difficult challenge for the pediatrician.

We report a rare case of a five-month-old female infant admitted in "Grigore Alexandrescu" Emergency Children's Hospital, Bucharest for diarrheic stools and severe malnutrition with birth onset. She was exclusively breastfed with poor gain weight (360g in 5 months). During the 5 months of life the child was repeatedly admitted to other hospitals because of diarrhea and failure to thrive without a precise diagnosis. We mention that she had a sister who died at 3 months old because of severe diarrhea with birth onset. At admission the infant had severe malnutrition with watery diarrhea and facial dysmorphism. After extensive investigations we excluded infections and allergic causes, cystic fibrosis, immune and autoimmune enteropathy, motility disorders. We performed inferior and superior endoscopies with biopsies that excluded microvillus inclusion disease. Examination of the biopsy specimens (optic and electronic microscopy) revealed the diagnosis of intestinal epithelial