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A RARE CASE OF SHWACHMAN-DIAMOND SYNDROME PRESENTING WITH DIABETES: IMPLICATIONS FOR PROGNOSIS

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Introduction. Shwachman-Diamond Syndrome (SDS) is a rare autosomal recessive genetic disorder characterised by pancreatic exocrine insufficiency (IP), bone marrow dysfunction as cyclic neutropenia, skeletal changes as metaphyseal dysostosis with short stature and a wide range of abnormalities and symptoms. After Cystic Fibrosis, SDS is the most common inherited cause of IP in the Western world.

Recently, a causative gene for this disorder containing five exons was identified at chromosome 7q11 (SBDS) and at least three mutations associated with SDS may be used to confirm diagnosis or for prenatal diagnosis. Clinical variations and specific changes age-related are common in this condition. A multidisciplinary strict monitoring could provide new insights into the course of SDS.

We report a rare case of SDS with diabetes mellitus (DM).

Case Report. G., a 15-year old boy, was diagnosed as SDS at 7 months according to diagnostic criteria of SDS including IP, bone marrow dysfunction, skeletal abnormalities, elevated serum aminotransferases, raised HbF. Normal sweat chloride concentration ruled out Cystic Fibrosis. He was admitted several times because of pneumonia and cyclic neutropenia. At 2 years of age he developed acute lymphatic leukaemia treated with allogenic bone marrow transplantation. During follow up metaphyseal dyschondroplasia was found on the bone radiograph.

In 2004, SDS was confirmed by performing genetic analysis which identified compound heterozygous for the most common mutations: 183–184TA>CT and 258+2T>C. The persisting growth retardation, despite an adequate pancreatic enzyme replacement and the rule out of GH deficiency, moved us to suspect the presence of DM without fasting hyperglycaemia. At the age of 12 an oral glucose tolerance test revealed a diabetic pattern of blood glucose levels (236 mg/dl at 120 min) with a poor insulin response (3.1 μ U/ml). Serum anti-insulin antibodies were not present. Neither metabolic acidosis or ketosis was found. Treatment with a long-acting insulin analog only partially controlled metabolic glucose pattern. Actually G. presents short stature, dental abnormalities, delayed puberty, growth retardation and behavioural difficulties.

Conclusions. Diabetes is a rare complication of SDS. Previous case reports show a wide range of clinical severity. Pathophysiology of developing diabetes remains no clear in SDS. Further studies are necessary to clarify the relationship between the SBDS gene and diabetes as well as the prognos-

tic role of DM. Currently short acting insulin is employed at each meal.

The development of national registers will help to elucidate clinical and genetic variations which correlate with better or worse prognosis in SDS.

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RECTAL PROLAPSE: AN UNUSUAL CASE OF FOOD ALLERGY IN A CHILD

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Introduction. Rectal prolapse in pediatrics has its highest incidence in infancy and is seldom seen in industrialised countries. The prolapse may involve only the mucosa (mucosal prolapse) or all layers of the rectum (complete prolapse or procidentia). It is sometimes detected by the child's parents and brought urgently to medical attention but has usually reduced spontaneously by the time they reach the practitioner's office. Rectal prolapse should be viewed as a symptom of an underlying condition rather than a separate illness. Potential causes are increased intra-abdominal pressure, diarrheal and neoplastic diseases, malnutrition, and conditions conducive to pelvic floor weakness. The strong association with cystic fibrosis makes the sweat test mandatory for infants and children with recurrent rectal prolapse.

Case report. The case described here regards a child aged 3 years 4 months, breast-fed for the first 3 months of life, with a clinical history of persistent constipation unaffected by treatment with laxatives and recurrent episodes of complete rectal prolapse after the introduction of cow's milk into his diet. The rectal prolapse disappeared after this substance was eliminated from the diet. There was no family history of atopy. Before admission for observation, the child had presented 8 episodes of rectal prolapse in the space of 3 years (the last 2 a fortnight before being admitted). While the results of prick tests carried out to ascertain whether the constipation was a symptom of food allergy proved negative, a patch test with cow's milk was positive. Total IgE were within the normal values and there were no specific IgE. The sweat test was negative. Colonoscopy revealed disorders of the mucosa compatible with the diagnosis of lymphoid hyperplasia, as confirmed by histologic examination of the biopsy specimens. This finding, which proves frequent in allergic enteropathies, suggested that an elimination diet for cow's milk protein might prove effective. The constipation disappeared over the following 6 weeks and there was no recurrence of rectal prolapse. When the patient was orally

exposed to fresh cow's milk as a check on the diagnosis, persistent constipation reappeared within 7 days together with complete rectal prolapse. The child was placed on a diet free of cow's milk protein. Follow-ups carried out after 2 and 6 months in the absence of pharmacological treatment confirmed the regularity of the intestinal function and the absence of rectal prolapse.

Conclusions. This case, the second reported in the literature, [1] appears to bear out the hypothesis that allergy to cow's milk protein can cause severe constipation combined with complete rectal prolapse through a delayed type of immunological mechanism, as suggested by the positive result of the patch test in the absence of immunological mechanisms of the reaginic type.

Reference

- [1] Shad A, Parikh D, Jawaheer G, Gornall P. Persistent rectal prolapse in children: sclerotherapy and surgical management. *Pediatr Surg Int* 2005;21:270–3.

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A CASE OF PNEUMOPATHY OF GASTROENTEROLOGIC INTEREST

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Case report. The case described regards a male child admitted for observation at the age of 9 years 9 months for persistent cough and disorders of esophageal motility. On birth, after a Caesarean section in the 33rd week for polydramnios, VATER syndrome was diagnosed due to the presence of esophageal atresia of the 3rd type, high anorectal malformation with rectovesical fistula, hypoplasia of the right kidney, agenesis of the coccyx, left hemivertebra of L3, dorsal butterfly vertebrae and accessory ribs. Three corrective operations were carried out at the ages of 3 days, 5 months, and 9 months: gastrostomy with positioning of a PEG tube, correction of the anorectal malformation by closing the rectovesical fistula, nephrectomy of the hypoplastic kidney and coloplasty of the esophagus to correct the esophageal atresia. Increase in weight was never satisfactory despite enteric feeding through PEG tube in the first ten months of life. Respiratory symptoms appeared at the age of 2 characterized by persistent cough and recurring pulmonary infections requiring 6–7 cycles of antibiotic treatment therapy a year. A marked degree of gastro-esophageal reflux was revealed by pH-metric examination and treatment with proton pump inhibitors proved only temporarily effective. Periods of moderate well-being alternated with the aggravation of symptoms. A marked worsening of the respiratory symptoms appeared at the age of 7 years 6 months with

increasingly persistent fits of coughing followed by the expulsion of undigested food from the oral cavity. At the time of the initial examination, an X-ray of the thorax showed a hypoexpanded right lung with basal confluent infiltrates and suppurating bronchiectasis with very marked pneumatoxis. The contralateral lung appeared emphysematous with a retrosternal mediastinal hernia. An X-ray of the digestive tube revealed inspissation of the proximal esophageal plicae and opacification of the transposed colic loop persisting for 7 h from the beginning of the examination due to total absence of peristalsis of the transposed section, which was emptied by gravity. There was also continuous contrast reflux from the stomach into the transposed section. A pulmonary CAT scan confirmed the bronchial and parenchymal disorders detected radiologically and revealed compression on the right principal bronchus by the ectatic neoesophagus with atelectatic phenomena resembling pneumonia ab-ingestis. Fibrobronchoscopy showed the presence of mucopurulent exudate in the right bronchial tree and esophagus gastroscopy revealed marked dilation of the transposed colon with the presence of alimentary material and broad continuity with the gastric cavity. The measures taken – respiratory physiotherapy, aerosol antibiotic treatment and proton pump inhibitors – led to moderate alleviation of the cough and a decrease in the frequency of alimentary vomiting. This comparative state of well-being continued after about 9 months of follow-up. As growth remained unsatisfactory (below the 3rd percentile for weight and height), PEG was carried out to improve the patient's nutritional state and the operation to replace the anastomotic tract was deferred to a later period, not least because the literature reports a high percentage of mortality in corrective reoperations.

Conclusions. Esophagocoloplasty must be the first choice for esophageal replacement in children. The complications are classified as minor (cervical anastomosis leak, abdominal evisceration, diarrhea, strictures, and reflux to the interposed viscera) and major (necrosis of transposed viscera, dehiscence of an intra-abdominal or intrapleural suture, torsion of transposed viscera, delayed gastric emptying reoperation, and cologastric anastomosis stricture). The interest of the case described here lies in the major complications of the respiratory apparatus closely related to the severe gastroesophageal reflux, which are very seldom reported in the literature.

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