Endocrine (2011) 40:146–147
DOI 10.1007/s12020-011-9460-7

LETTER TO THE EDITOR

A rare case: Shwachman–Diamond syndrome presenting with diabetic ketoacidosis

M. Fatih Akdogan · Mustafa Altay · Nazim Denizli · Murat Gucun · Seher Tanrikulu · Murat Duranay

Published online: 30 March 2011

Dear Sir,

Shwachman–Diamond syndrome (SDS) is a rare autosomal recessive multi-system disease that usually manifested with exocrine pancreatic insufficiency, short stature, and chronic neutropenia [1]. Previously only a few SDS patients have been reported to develop diabetes [2]. We report a case of SDS with diabetic ketoacidosis and osteopenia in which mutations of the SDS gene have been identified.

A 29 year old woman was admitted to emergency room with symptoms of frequent urination and confusion. For the last 15 days, her oral intake was not satisfactory because she had nausea and vomiting. She had no history of diabetes mellitus or any other disease. On her examination, she had no orientation and cooperation. She appeared dehydrated and her mouth and mucosas were dry. Lab findings were as follows: Hg: 15.5/μl, WBC: 1200/μl (NEU: 100/μl), glucose: 483 mg/dl, BUN: 25 mg/dl, creatinine: 1.46 mg/dl, potassium: 1.7 mEq/l, total protein: 5.5 g/dl, albumin: 2.2 g/dl, calcium: 5.6 mg/dl, phosphorus: 0.1 mg/dl, pH: 6.94, and HCO_3: 4.3 mmol/l. At urinalysis, ketonuria was +3 and glucosuria was +4.

Her clinical and laboratory findings were consistent with diabetic ketoacidosis. Patient was treated with iv fluid and electrolyte replacement and insulin infusion. Her body mass index was calculated as 27.5 kg/m^2. 25 OH vit D was 6 ng/ml and PTH was 99 pg/ml. Her HbA1c was 11.4%, C-peptide was 1.8 (0.4–4), and anti-insulin antibody was positive.

After these findings, the patient was investigated for malnutrition. Stool analysis revealed rare protein fibers. Her serum amylase and lipase were determined at relatively lower levels. Because she had hypocalcemia and hypophosphatemia, DEXA was performed and osteopenia (L2 *T* score: −1.65, *Z* score: −1.63 and trochanter of femur *T* score: −2.29, *Z* score: −1.95) was detected. Pancreatic atrophy was detected on abdominal computed tomography. Genetic analysis showed monosomy 7. So the patient was diagnosed as SDS. Pancreatic enzymes and insulin replacement was administered. After clinical and laboratory improvement, she was discharged and she had cyclic neutropenia attacks in her follow up.

In the interesting case, because she had neutropenia, findings of malabsorption and pancreatic atrophy, the patient was considered as SDS. Genetic analysis for monosomy 7 confessed the diagnosis. SDS is usually associated with insufficiency of exocrine but not endocrine functions of pancreas. Only a few cases have been reported about diabetes developed in SDS patients before. Coexistence of SDS and diabetes is unlikely to be incidental, the onset and severity of diabetes varies with each patient, and the pathophysiology of developing diabetes remains unclear [3].

Osteoporosis and SDS had been assessed in a few studies in literature. SDS osteoporosis is characterized by reduced numbers of osteoclasts and osteoblasts and low bone turnover and may be directly related to the bone marrow dysfunction and neutropenia. Malabsorption could be an additional risk factor [4]. Rosendahl et al. [5] reported an adult SDS patient with DM and osteoporosis

M. Fatih Akdogan
Department of Internal Medicine IV, Haydarpasa Numune Education and Research Hospital, Istanbul, Turkey

M. Altay (✉)
Department of Endocrinology, Medical Faculty, Gazi University, Besevler, Ankara, Turkey
e-mail: altay_mustafa@hotmail.com

N. Denizli · M. Gucun · S. Tanrikulu · M. Duranay
Department of Nephrology, Haydarpasa Numune Education and Research Hospital, Istanbul, Turkey

such as this case. Differently, the patients' osteodensity scores were slightly worsened (osteopenia).

In conclusion, despite diabetes mellitus and SDS rarely coexists, the probability of this condition must be considered in diabetic patients whose clinical features fit. Genetic analysis should be done to support diagnosis and patients should be followed up to late stages in order not to overlook complications such as osteoporosis and malnutrition.

Conflicts of interest The authors declare that they have no conflict of interest.

References

1. P.J. Aggett, N.P. Cavanagh, D.J. Matthew, J.R. Pincott, J. Sutcliffe, J.T. Harries, Shwachman's syndrome: a review of 21 cases. Arch. Dis. Child. **55**, 331–347 (1980)
2. D.H. Shmerling, A. Prader, W.H. Hitzig, A. Giedion, B. Hadorn, M. Kuhni, The syndrome of exocrine pancreatic insufficiency, neutropenia, metaphyseal dysostosis and dwarfism. Helv. Paediatr. Acta **24**, 547–575 (1969)
3. L. Filippi, M. Tronchin, M. Pezzati, G. Chiti, C. Dani, G.F. Vichi, F.F. Rubaltelli, Shwachman syndrome in a preterm newborn associated with transient diabetes mellitus. J. Pediatr. Gastroenterol. Nutr. **34**, 219–223 (2002)
4. S. Toiviainen-Salo, M.K. Mäyränpää, P.R. Durie, N. Richards, M. Grynpas, L. Ellis, S. Ikegawa, W.G. Cole, J. Rommens, E. Marttinen, E. Salivahti, O. Makitie, Shwachman–Diamond syndrome is associated with low-turnover osteoporosis. Bone **41**, 965–972 (2007)
5. J. Rosendahl, N. Teich, J. Mossner, J. Edelmann, C.A. Koch, Compound heterozygous mutations of the SBDS gene in a patient with Shwachman–Diamond syndrome, type 1 diabetes mellitus and osteoporosis. Pancreatology **6**, 549–554 (2006)