

A PATIENT WITH SECKEL SYNDROME ASSOCIATED WITH PARTIAL IgA DEFICIENCY AND IDIOPATHIC THROMBOCYTOPENIC PURPURA

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ÖZET

PARSİYAL İGA EKSİKLİĞİ VE İDİOPATİK TROMBOSİTOPENİK PURPURA İLE BİRLİKTE GÖRÜLEN SECKEL SENDROMU

Bu çalışmada 5½ yaşında idiyopatik trombositopenik purpura ve parsiyel İgA eksikliği ile birlikteliği olan Seckel sendromu sunuldu. Hastada alt ve üst solunum yolu, gastrointestinal sistem ve üriner sistemi içeren tekrarlayıcı enfeksiyonları vardı. Hastada 5 yaşında idiyopatik trombositopenik purpura ortaya çıktı. Seckel sendromlu hastada tekrar eden enfeksiyonların görülmesi halinde immün yetmezlik araştırılmalıdır.

Anahtar terimler: Seckel sendromu, İgA eksikliği, idiyopatik trombositopenik purpura.

SUMMARY

A 5½-year-old girl with Seckel syndrome associated with partial IgA deficiency and idiopathic thrombocytopenic purpura has been presented. The patient experienced recurrent infections including upper and lower respiratory tract, gastrointestinal and urinary systems. She had also idiopathic thrombocytopenic purpura at the age of five. Patient with Seckel syndrome presenting frequent infections should be evaluated for immunodeficiencies.

Key words: Seckel syndrome, IgA deficiency, idiopathic thrombocytopenic purpura.

INTRODUCTION

Seckel syndrome that was described by Seckel (1) in 1960 is a rare autosomal recessive inherited malformation. Major features of this syndrome are bird-headed dwarfism, prenatal, postnatal microcephaly and growth retardation, a typical face, mental deficiency and other minor deformities (1,2). The patients are usually pleasant with the sociable personality. We describe here a patient with Seckel syndrome associated with partial IgA deficiency.

CASE REPORT

A 1½-month-old girl was admitted with complaint of failure to thrive in 1990. Her parents who are consanguineous (first cousins) have three other healthy daughters. The patient was born at term with the weight 1600 g, height 43 cm and head circumference 29 cm respectively. Physical examination revealed microcephaly, aged appearance, wide eyes, prominent beaked nose, low set ears, micrognathia. She was diagnosed as Seckel Syndrome.

The patient has been followed up in our clinic and is now 5½-years-old with the following findings; microcephaly (head circumference 30 cm), short stature (87 cm), mental retardation and scoliosis (Fig 1).

So far, the patient had experienced recurrent infections, including five times broncopneumonia, three times gastroenteritis and dehydration caused by aemibiasis and giardiasis, three times urinary infections and at least four episodes of upper respiratory infections a year. She also had purpuric lesions and epistaxis due to idiopathic thrombocytopenic purpura (ITP) at the age of five. Hematological investigation revealed hemoglobin 13.5 g/dl, white blood cell count 5700/mm³ with normal distribution and platelet count 54.000/mm³. Bone marrow aspiration was normal with increased number of megacaryocytes. The disease recovered within two months. Laboratory and radiological examinations including ultrasonography, intravenous urography and dimercaptosuccinic acid (DMSA) scan showed no other system abnormalities but low level of serum IgA. Serum IgA levels were lower than two standard deviation of age matched healthy population in consequent measurements at different ages and did not raise with age (3). Current serum IgG, IgM and IgA levels of the patient measured by nephelometer (Beckman Protein Array System, Beckman Inc. USA) were 9.68 g/L, 0.99 g/L, and 0.46 g/L respectively.

DISCUSSION

Seckel syndrome is considered to be an autosomal recessive disorder with the incidence less than 1:10.000 (1). The first degree of consanguinity of the parents (first cousins) suggests also autosomal recessive transmission in our patient. This patient corresponds well with the general description of Seckel syndrome. Several other manifestations, such as cardiac anomalies, hematological disorders were reported in patients with Seckel syndrome (4,5). Butler et al (6) reported Fanconi anemia in a two cases with Seckel syndrome. Hayani et al (7) showed acute myeloid leukemia in a case. In these two reports chromosomal abnormalities were also detected (6,7). In our patient the major noticeable features were the partial IgA deficiency and ITP. IgA deficiency is characterized by its clinical

variability associated with recurrent infections, allergic, hematological, autoimmune diseases and malignancies (8,9). We suppose that partial IgA deficiency explains the occurrence of recurrent infections and ITP. We were not able to find any other report subjecting Seckel syndrome associated with immunodeficiencies. On the other hand, with additional or different osteodysplastic features, Seckel-like syndrome



Figure 1. Aged appearance, wide eyes, prominent beaked nose, low set eyes, micrognathia, microcephaly at the age of 5 1/2 years old.

was also described (10,11). There were no such findings in this patient.

We followed the mother by serial ultrasonography in subsequent pregnancy for the prenatal diagnosis of Seckel syndrome. No abnormality was detected and the mother gave birth to a healthy another girl.

It would be suggested that the patients with Seckel syndrome experiencing recurrent infections and autoimmune disorders should be investigated for immunodeficiencies in order to clarify the true incidence.

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