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Rothmund Thompson Syndrome: A Case Report Presenting with Aspiration Pneumonia

Rotmund Thompson Sendromu: Aspirasyon Pnömonisi ile Prezente Olan Bir Olgu

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Abstract

In the few studies of Rothmund Thompson Syndrome (RTS) to date in literature, two clinical subforms have been identified: the RTS-I form, characterized by poikiloderma, ectodermal dysplasia and juvenile cataract; and the RTS-II form, characterized by congenital bone defects, increased risk of childhood osteosarcoma and skin cancer. We present here a case with the RTS-II form with bone deformities, history of skin cancer and growth retardation. The patient was admitted to our hospital with complaints of dyspnea for two days. Total atelectasis in the right lung and severe dilatation and obstruction in the distal esophagus were observed on a posteroanterior chest X-ray and thorax computed tomography, and antibiotherapy, oxygen therapy and supportive treatments were initiated after a diagnosis of aspiration pneumonia was made. Alveolar hemorrhage and hematuria developed during follow up due to bleeding disorder. Despite supportive treatment, the patient developed multiple organ failure (respiratory failure, renal failure, pancytopenia), and died on the 8th day of hospitalization. We present this case study to draw attention to the risk of bleeding tendency and aspiration pneumonia in RTS patients.

Key words: *Rothmund Thompson Syndrome, Aspiration Pneumonia, Esophageal Dilatation, Hemorrhage.*

Öz

Rothmund Thompson Sendromu (RTS) oldukça nadir görülen bir sendromdur. İki klinik alt formu tanımlanmıştır: poikiloderma, ektodermal displazi ve jüvenil katarakt ile karakterize RTS-I formu ve konjenital kemik defektleri, çocuklukta artmış osteosarkom ve cilt kanseri riski ile karakterize RTS-II formudur. Burada sunulan hasta, kemik deformiteleri, cilt kanseri öyküsü ve gelişme geriliği nedeniyle RTS-II formu idi. Hasta, iki gündür başlayan nefes darlığı ve hırıltı şikayeti ile hastanemize başvurdu. Posteroanterior akciğer grafisinde ve toraks bilgisayarlı tomografide sağ akciğerde total atelektazi, özefagusda ileri düzeyde dilatasyon ve özefagus distalinde obstrüksiyon izlendi. Aspirasyon pnömonisi tanısıyla, antibiyoterapi, oksijen tedavisi ve destek tedavi başlandı. Tedavi altında iken, kanama bozukluğu nedeniyle alveolar hemoraji ve hematüri gelişti. Destek tedaviye rağmen çoklu organ yetmezliği (solunum yetmezliği, böbrek yetmezliği, pansitopeni) gelişen hasta 8. günde exitus oldu. Olgu RTS hastalarında kanamaya eğilim ve aspirasyon pnömonisi gelişimine dikkat çekmek adına sunulmuştur.

Anahtar Sözcükler: *Rotmund Thompson Sendromu, Aspi¸rasyon Pnömonisi¸, Özefagus Dilatasyonu, Hemoraji.*

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Rothmund Thompson Syndrome is a very rare disease that has been reported on in 300 patients in literature (1). It was first described in 1868 by ophthalmologist Rothmund who observed growth retardation and rapidly progressive bilateral juvenile cataracts and poikiloderma in 10 children in a Bavarian village. In 1936, British dermatologist Thomson reported poikiloderma, skeletal defects, growth retardation in three children, but no cataracts. We present here as case of RTS with bleeding tendency who was diagnosed with aspiration pneumonia.

CASE

A 29-year-old female patient was admitted to our hospital with complaints of dyspnea after food intake. The patient had diagnosed hypothyroidism, esophageal dilatation and RTS, and a history of surgery to the right ankle due to skin cancer in childhood, and had undergone splenectomy and femoral fracture surgery three years earlier. A physical examination revealed mental confusion, decreased respiratory sounds in the right lung and sonor crackles in the left lung. Pulse 02 saturation was 70% and heart rate was 120/min. Radiopacity was observed due to total atelectasis in the right lung on a chest X-ray (Figure 1). Total atelectasis was observed in the right lung, the esophagus was noted to be severely dilated and the distal esophagus to be obstructed on thorax CT (Figure 2). Piperacillin tazobactam antibiotherapy, prophylactic dose anticoagulant, deep tracheal aspiration and respiratory physiotherapy were initiated with a diagnosis of aspiration pneumonia. Hematuria developed following the insertion of a catheter. The hemoglobin blood level was followed up and continuous irrigation was applied upon the recommendation of urology. The patient was intubated due to the lack of clinical or radiological improvement despite antibiotherapy and supportive treatment. After intubation, regression of the right atelectasis was noted on a control chest X-Ray (Figure 3), however, diffuse alveolar hemorrhage developed due to mechanical cleaning due to bleeding tendency (Figure 4). The patient received two units of fresh frozen plasma and three units of erythrocyte suspension due to pancytopenia and alveolar hemorrhage. Despite supportive treatment, multiple organ failure (respiratory failure, renal failure, pancytopenia) occurred and the patient died on the 8th day of hospitalization.

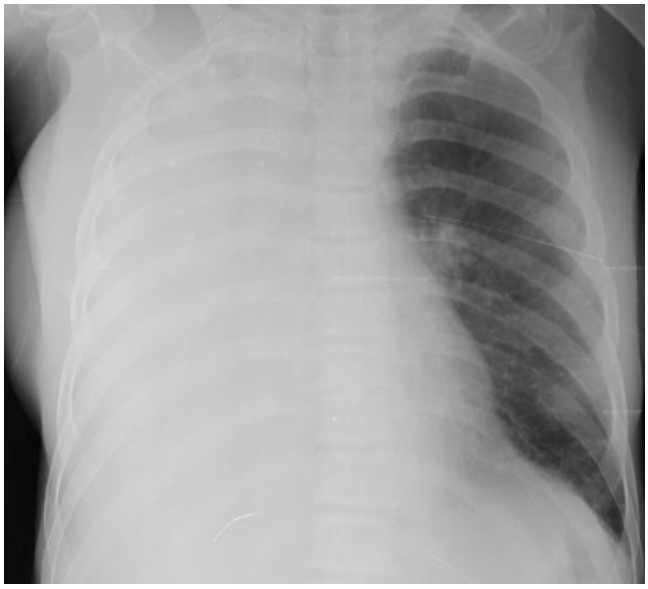

Figure 1: Total atelectasis of the right lung revealed on the initial chest X-Ray upon hospital admission

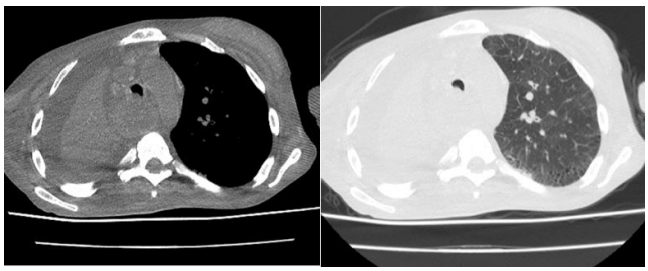

Figure 2: Atelectasis in the right lung and dilatation of the esophagus seen on Thorax CT

DISCUSSION

Two clinical subforms of RTS have been described in literature: the RTS-I form, characterized by poikiloderma, ectodermal dysplasia and juvenile cataracts; and the RTS-II form, characterized by congenital bone defects, an increased risk of osteosarcoma in childhood and skin cancer in adult life. While the RTS-II form is caused by homozygous or compound heterozygous mutations in the RECQL4 helicase gene, detected in 60–65% of RTS patients, the etiology of RTS-I is unknown (2). RECQ proteins are conserved DNA strands that act as helicases, and the RECQ helicase family plays a role in the regulation of aging while also acting as tumor suppressors. Mutations in RECQ4, one of the RECQ family proteins, therefore, not only result in developmental abnormalities and cancer predispositions, but also potentially premature aging. Patients with RTS-II are thus at risk of both advanced age cancer and signs of premature aging in the skin (3).

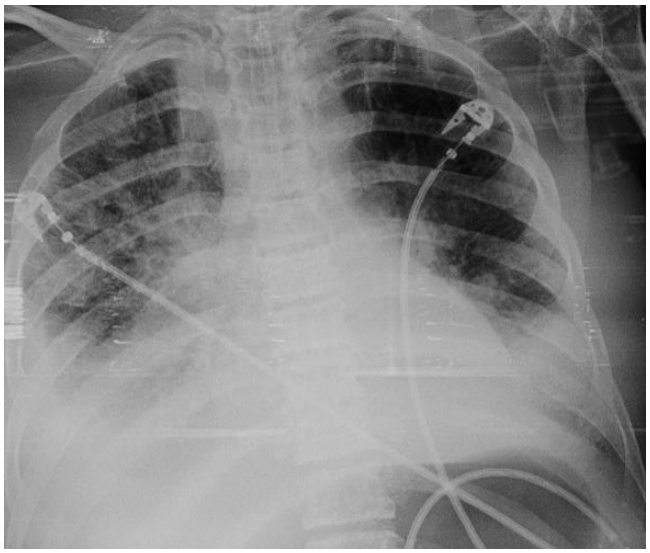

Figure 3: Recovery of atelectasis seen in the right lung one day after deep tracheal aspiration

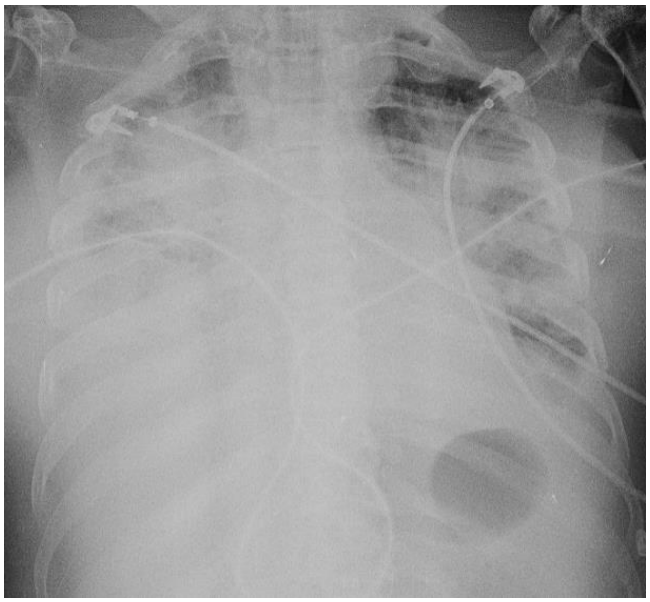

Figure 4: Alveolar hemorrhage seen on chest X-Ray after massive hemoptysis on day 2 following intubation

The case we present here had the RTS-II form of the condition. Concurring with previous studies in literature, the patient was mentally normal, had sparse thin hair, a dysmorphic jaw and tooth structure, microdontia, short stature and low weight, keratosis and sclerosis of the skin, saddle nose and esophageal dilatation due to pyloric stenosis, as detected on physical examination.

Previous studies have described progressive leukopenia requiring transfusion, chronic microcytic hypochromic anemia, malignant hematological abnormalities ranging from myelodysplasia to aplastic anemia and leukemia in RTS cases (4,5). In our patient, erythrocyte suspension and fresh frozen plasma replacement were applied to the treatment protocol due to microcytic anemia, leukopenia and low platelet. Subcutaneous vasodilation (telangiectasia) and hematological abnormalities are common in RTS patients, making them prone to bleeding during interventional procedures. In our patient, a bleeding tendency was observed after all kinds of interventional procedures, such as catheter insertion and deep tracheal aspiration.

Bronchiectasis and recurrent pneumonia have been described only rarely in patients with RTS (4,6,7). Although bronchiectasis was not observed in our case, there was total atelectasis due to aspiration pneumonia in the right lung.

CONCLUSION

This case emphasizes that gastrointestinal pathologies are common in RTS patients, and that aspiration pneumonia may develop after esophageal dilatation. The study further draws attention to the occurrence of hemoptysis and hematuria due to the likelihood of bleeding from mucosal tissue.

CONFLICTS OF INTEREST

None declared.

AUTHOR CONTRIBUTIONS

Concept - H.İ.Y., H.I.K., G.A., A.C.P., H.K., M.P.; Planning and Design - H.I.K., H.İ.Y., M.P., A.C.P., H.K., G.A.; Supervision - A.C.P., G.A., H.K., M.P., H.İ.Y., H.I.K.; Funding - M.P., H.K., H.İ.Y., G.A., A.C.P.,H.I.K.; Materials - H.İ.Y., H.I.K., A.C.P., G.A., H.K., M.P.; Data Collection and/or Processing - M.P., H.K., H.İ.Y., H.I.K., A.C.P., G.A.; Analysis and/or Interpretation - A.C.P., G.A., H.İ.Y., H.I.K., M.P., H.K.; Literature Review - M.P., H.İ.Y., H.K., H.I.K., A.C.P., G.A.; D.A.; Writing - H.İ.Y., H.I.K., A.C.P., G.A., M.P., H.K.; Critical Review - A.C.P., G.A., H.K., H.İ.Y., H.I.K., M.P.

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