American Journal of Medical Case Reports, 2015, Vol. 3, No. 2, 30-31
Available online at http://pubs.sciepub.com/ajmcr/3/2/2

DOI:10.12691/ajmcr-3-2-2

Fever of Unknown Origin with Psychiatric Complaints in a Patient with Rubinstein-Taybi Syndrome

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Received November 19, 2014; Revised January 17, 2015; Accepted January 19, 2015

Abstract Rubinstein–Taybi syndrome (RTS) is a rare genetic disorder characterized by mental retardation, postnatal growth deficiency, microcephaly, specific facial characteristics, broad thumbs, and big toes. Presence of dental problems and inadequate follow-up in RTS, as well as difficulty in self-care, poses a substantial health threat. This case report describes the evaluation of a 27-year-old female RTS patient diagnosed with fever of unknown origin (FUO) with psychiatric complaints and underlines the importance of physical examination including oral cavity in patients with difficulties in expressing their physical complaints.

Keywords: *Rubinstein-Taybi Syndrome, oral care, fever of unknown origin, mental retardation*

Cite This Article: İbrahim Taymur, Buket Belkız Güngör, Cenk Aypak, Ahmet İnel, and Özgür Dağlı, "Fever of Unknown Origin with Psychiatric Complaints in a Patient with Rubinstein-Taybi Syndrome." *American Journal of Medical Case Reports*, vol. 3, no. 2 (2015): 30-31. doi: 10.12691/ajmcr-3-2-2.

1. Introduction

Rubinstein-Taybi Syndrome (RTS), or Broad Thumb-Hallux syndrome, includes a group of congenital anomalies consisting of short, broad thumbs and great toes, psychomotor retardation, highly arched palates, and particular facial abnormalities. [1] It is caused by either a microdeletion at 16p13.3 or mutations in the CREB-binding protein (CREBBP or CBP) or EP300 gene (at 22q13). [2]

Oral manifestations of this syndrome include limited mouth opening, a pouting lower lip, retro/micrognathia, a high arched and narrow palate, cleft uvula and palate, and rarely a cleft upper lip. Dental abnormalities occur in 67% of individuals with RTS and can include hypodontia, maintenance of deciduous teeth, talon cusps, and enamel hypoplasia. An increased rate of caries and periodontal disease has been reported in these patients [2,3].

Patients with psychomotor retardation as seen in RTS have difficulty in expressing their complaints or conditions related to their self-care. [2,3]. Here we report a 27-year-old female RTS patient diagnosed with fever of unknown origin (FUO) with psychiatric complaints in order to underline the importance of physical examination including oral cavity in patients with difficulties in expressing their physical complaints.

2. Case Report

A 27-year-old female patient was being followed and treated for RTS and moderate mental retardation. Aggressive behaviors, impaired impulse control, insomnia, unstable mood, talking to herself, visual hallucination and agitation complaints appeared when she was 15 years old and significantly disappeared after receiving 500 mg/day divalproex therapy. She had high fever, difficulty in speaking and walking, nausea, shivering, agitation attacks, fatigue, and weakness 5 months ago; these complaints have worsened in a few days. She had been admitted to a tertiary hospital and hospitalized at infectious disease clinic to be investigated for the etiology of fever, which reached 39°C (axillary). Over the course of 2-month hospital stay, she underwent detailed examination including routine biochemistry, cranial MRI, whole abdominal US, abdomen CT, blood and urine culture, peripheral blood smear, and brucella agglutination testing; but continuing high fever and clinical complaints persisted and she was discharged with the diagnosis of "fever of unknown origin" (FUO). Since her clinical symptoms did not improved and she additionally developed psychiatric symptoms (agitation attacks, insomnia and continuous shouting), the patient was admitted to psychiatry clinic. Her mental examination at admittance to psychiatry clinic revealed that she was conscious and her orientation were normal, previous friendly communication was impaired and she had difficulty in speaking, she was agitated and unable to establish communication, unable to walk, and had been using wheel chair for the last 3 months. Treatment of the patient, who had been receiving 500 mg/day divalproex therapy until hospitalization, was switched to 2 mg/day risperidone, 2 mg/day lorazepam and 40 mg/day propranolol on the first day of her hospitalization. No significant improvement was observed in her overall medical condition and psychiatric

complaints within the first week of hospital stay. Considering potential tooth decay on routine examination, she was consulted with the dentist in the hospital. On the 10th day of hospitalization, oral examination revealed tooth decay and she underwent tooth extraction under anesthesia. Five days after tooth extraction, it was observed that high fever dramatically disappeared, she began walking, and her agitation and screaming attacks notably decreased. Considering the clinical improvement on the 2nd week of hospitalization, her medical treatment was switched to 1mg/day lorazepam and 20 mg/day propranolol. Her general wellbeing was notable on the 20th day of hospitalization and she was discharged. On the second month of her follow-up all her complaints disappeared. She has been continuing private education, and has gained previous quality of life back.

3. Review of Literature & Discussion

Behavioral problems such as impulsivity, mental retardation, unstable mood, stereotype, inclination to irritability, friendly and rapid social contact, short-term attention, and difficulty in locomotor functions are frequently encountered in the patients with RTS [4,5]. Cases with mood disorders (mania) and psychosis have been also rarely reported [6,7]. It has been stated that psychiatric symptoms and disorders observed in RTS patients might arise from the relation between 16q13 chromosome deletion and abnormalities of gamma-aminobutyric acid (GABA) receptor or neurotransmitter. In the literature, in a RSTS case that had mania, it was demonstrated that divalproex therapy notably improved the complaints after treatment. Likewise, in the present study, divalproex therapy performed well for mania and similar behavioral disorders, which appeared when the patient was 15 years old, was considered beneficial and thereby psychiatric follow-up and treatment was carried out. Presence of mental retardation and additional psychiatric disorders spoiled both functionality and self-care also in the present case and caused caregivers to have difficulties in nursing the patient.

Presence of considerable dental problems and inadequate follow-up in the patients with mental retardation, as well as difficulty in self-care, poses a substantial health care problem [8]. RSTS is a rarely encountered mental retardation syndrome and brings along increased risk of dental disease-related infection. Dental problems such as multiple dental anomalies, periodental diseases, severe malocclusion, decreased care for oral hygiene, and tooth decays are frequently encountered in the patients with RSTS. Nevertheless, evaluation of 45 RSTS patients revealed oral appearance defined as thin upper lip, small oral opening, pouted lower lip, retro/micrognathia, and apparently higher-arched narrow palate. [9] Prevalence of tooth decay is increased by 62%, and after long-term and cross-sectional evaluation of these patients, it was stated that periodic oral care is important [9].

In the present case, infection due to tooth decay caused high fever, impairment in general status, and increase in psychiatric complaints, and all her complaints dramatically improved in a short time after treatment of tooth decays. Fever of unknown origin is an important health problem for clinicians. There might be cases in which no result could be obtained in spite of recent advanced technologies used to detect etiology of fever. Dental diseases are found to be as a potential cause in such cases [10]. In the present case, high fever persisted for 5 months due to tooth decay. In the literature, similarly, a case with high fever that persisted for 2 years due to dental abscess was reported [11].

4. Final Comments

General physical examination gains more importance as the patients with psychiatric diseases have difficulty in expressing their complaints. Oral and dental examination is one of the most important components of physical examination. The present case emphasizes the importance of oral care and hygiene in the patients with impaired cognitive functions.

On behalf of all authors, the corresponding author states that there is no conflict of interest.

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