Which type of neurofibromatosis? A report about a rare case

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Abstract

Introduction: Neurofibromatosis is a genetic disease that is found in two separated types with different prevalences. In various societies and with attention to both types of Neurofibromatosis, they divulge their proper clinical remarks separately. Presence of mentioned remarks in the patients is of interesting points that available conditions are seen rarely in affected persons to Neurofibromatosis.

Case report: The patient was a 26-year-old boy who seemed younger. He suffered from severe hiccups since three days ago, tension headache in frontal region, nausea and elastic sudden vomiting. Other important clinical points in the patient was the presence of skin wastage of Neurofibromatosis in six separate sites with approximate dimension of 7 to 8 cm, café au lait from the time he was 3 years old, cardiac tumor in dimensions of 1×2 cm in right auricle and a pile in dimension of 7cm at right parietal lobe.

Discussion: According to the patients biography, presence of mentioned remarks and progressive process of disease which culminate to death four days after diagnosis, so pathogenesis and process of disease fulminant and death of patients clearly were not distinguished and it is seemed that disciple of recent reports about mentioned new types on the subject of Neurofibromatosis. This instance also should be under consideration on title of new type of Neurofibromatosis.

Keywords: **neurofibromatosis, café-au-lait lesions**

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Introduction

First type of neurofibromatosis or von recklinghausen disease, is one of known disorders in genetics with a low prevalence (one in 3000 birth)[1] that appears because of mutation in long arm of Chromosome number 17 and inherited in the shape of prevailing autosomal.[2,3] This disease was defined and explained completely for the first time in 1882 by a person named Von recklinghausen.[4] The incidence of the second type of neurofibromatosis is much less than the first one (one in 55000 birth),and it was defined for the first time in 1930.This disease appears because of disorder in chromosome number 22,and it was not considered as a separated type of disease still 1981(the first and the second type were considered as one disease).[5,6] The first type has some known symptoms such as café-au-lait lesions, tumors of nerve system, and ocular complication. There is no café-au-lait lesions in the second type of neurofibromatosis, but it appears with CNS tumors, acousticus neurinom complications, and hearing lost. The important point in our patient was the presence of symptoms related to both types of neurofibromatosis. Available conditions just seen in a rare patients of neurofibromatosis, in a way that these patients have café-au-lait lesions, dysparietal region one-side tumor, symptoms of low-hearing, and long hiccups resisted against treatment that probably occurs because of involvement of brain nerves number 9 &10.Another important point in this case is severe respiratory distress and echocardiography reported right atrium hypertrophy because of heart tumor with the size of 1×2 cm.

Case report

Patient was 26-years-old, but looked as a 15 years old boy. He suffered from severe hiccups for three days, headache in frontal region, and elastic sudden vomiting that lasted about one month with progressive regression trend. Our patient lost 10 kilograms in the last week of his life-time and complained of hiccups that lead to difficulty in talking, low hearing, and severe polyuria (20 times during 24 hours).He was mentally safe ,did computing well, and also had good behavior in the society but his difficulty in hearing was obvious. His parents mentioned measles at the age of three and after that, café-au-lait lesions (figure 1) and skin tumoral lesions(figure 2) appeared in his body and prevented him to check about café-au-lait lesions.

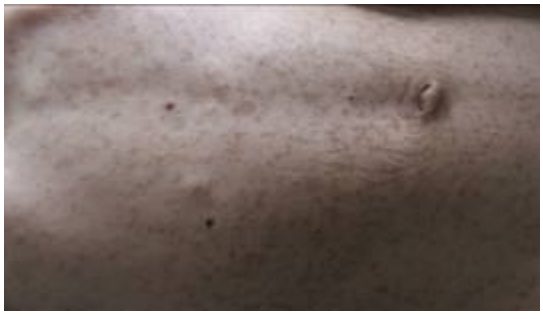

Figure 1

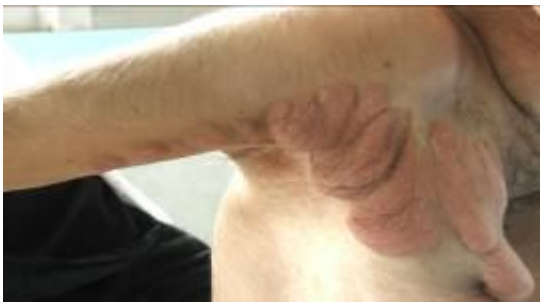

Figure 2

In his examination we understood that these lesions spread through all of his body and there were well-defined soft tumoral lesions in the shape of multiplex neurofibromatosis in six zones of his body. Severe chest deformity in the sternum zone was toward inside without any respiratory restriction. In the head and face exam, just café-au-lait lesions were seen. In the heart auscultation a kind of week click was hearing that by echocardiography sternum (vidit 4)was suggested because of cardiac tumor with the size of 1×2. Lung auscultation was clear; the abdomen was fine without any tenderness on organomegaly. Left limbs forces decreased without any deformity, his genital organ matched with his real age not with his appearance. Systolic blood pressure verified from 100 to 140 mmhg. Our patient had no fever and there were no multiple spinal anomalies, kyphoscolyosis and degeneration of lumbar lordosis (figure 3). A mass with the size of 7×8 was seen in the chest X-ray in brain CTscan in right partial lobe (figure4) that caused shifting of nearby parts to other side.

Figure 3

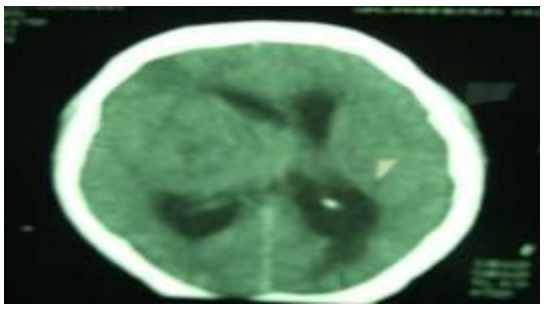

Figure 4

Discussion and conclusion

In the inspections and examinations, our patient complained from long headaches that resisted a lot against drugs. In MRI with contrast, the brain tumor was identified as the origin of his headache. He also suffered from vertigo and hearing loss that with the request of audiometry and sensory neural type of hearing loss and also according to the observed brain tumor, acousticus neurinom was specified. Neurofibromatosis appeared with various expressions which oropharynx,

soft tissue of neck, and mediastinum among them have special importance.[8,9]

However several different studies show that the main reasons of 24 percent of deaths caused by severe neurofibromatosis are malignancies.[7]

In the examination of patients, plexiform neurofibromatosis skin lesions with the size of 7 to 8 cm were seen in the separate six zones. These lesions that are known as Bag of Worms (figure 5) appeared with thick and changed skin.

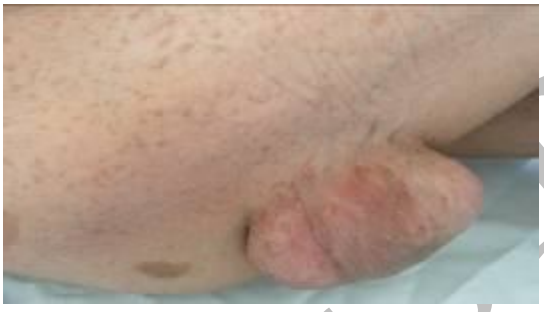

Figure 5

Plexiform neurofibromatosis lesions are seldom painful and most of the times the pain in the lesions is the sign of malignant peripheral nerve sheath tumors (MPNST).[1,48] The patient suffered from the appearance of pain in the lesions at the end of consciousness period that it was for the wild spread malignant in his brain and also because of complicated MPNST. MPNST were more severe in neurofibromatosis patients.[49,50] MPNST patient's prognoses depends on several factors. The tumor of over 5cm. size and high histological grade are rare factors.[49,50,54,55] But they consider as poor prognosis that in our case such factors affected on prognosis. Although surgery manner and tumor excision are effective in the survival determination of these patients,[49-53,55] but we couldn't use them because of bad condition. Abdominal pain, one of the important clinical signs probably was because of generalized dilatation of bowel loop. Today gastrointestinal features of neurofibromatosis are well-known in a way that it was in the previous similar studies.[10-12] In 11 to 25 percent of these patients, gastrointestinal complication was reported. These features might be mistakenly known as Hirschprung[13,14] in children and Chronic pseudo-obstraction and megacolon in adults.[15,16]

Stroma tumors are the most common GI tumors in the neurofibromatosis that often can be found in the stomach and duodenum. These lesions typically reported with the obstruction, hemorrhage, intussusceptions, volvolus, and perforation.[10,17-20] Although GI lesions in these patients are often benign but in several cases, GI malignancy such as Leiomyosarcoma and malignant schwannoma are reported [20-22]. At the

end it was found that there is obvious relationship among incidence of neurofibromatosis, duodenal carcinoid tumors, and pre-ampulatory.[23-25]

Although there was no founding of duodenal carcinoid tumors but of the other interesting points in our case was systolic blood pressure (BP) of 140 mmHg.

According to the previous founding of systolic BP of 80, the sudden raising of BP can be one of the other sign of intensification of neurofibromatosis complications that occurs secondary and as a result of kidney vessels narrowing that in 25 percent reports accompanied by abdominal Coarctation of the aorta.[26-28] Although in the sonograghy of our case, the abdominal aort or regional arteries coartation sign were not found, but it is important to mention that hypertension of patient may be for the central disorder of the tumor. Although kidney artries, vertebral artery , subclavian, and aorta aneurysm are of the vessels sign in this disease,[29-36] in many cases massive bleeding occurs as a result of vessels disorders that caused from general surgery.[29, 32-34] Vessels involvement are very important because they are the common cause of the hypertension in children with neurofibromatosis, Coarctation of the aorta and kidney vessels disorders. Dyspnea and dysphagia are of the other cardiothoracic features in our patient that can be with vagus involvement appears and by creating of disorder in left side of thorax causes sudden death. Also thoracic bleeding resulted from subclavian vessels ruptures and internal mamilary artery with unknown origin reported frequently.[42-47] Finally according to the patient's history and symptoms of two types of neurofibromatosis and progressiveness of the disease, our patient dead after four days. Pathogens of the disease and the reason of death were not completely cleared.

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کدام تیپ از نوروفیبروماتوز؟ - گزارش یک مورد نادر

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چکیده

مقدمه: نوروفیبروماتوز یک بیماری ژنتیکی می باشد که بصورت دو تیپ مجزای یک و دو و با شیوع متفاوت در جوامع مختلف دیده می شود با توجه به اینکه هر دو نوع تیپ نوروفیبروماتوز به طور مجزائی علائم بالینی خاص خود را بروز می دهند وجود علائم مربوط به هر دو نوع تیپ نوروفیبروماتوز از نکات جالب و قابل توجه در بیمار مورد نظر به شمار می آید که شرایط موجود تنها در شیوع بسیار بسیار کمی از مبتلایان به نوروفیبروماتوز دیده می‌شود.

معرفی بیمار: بیمار پسری با سن تقویمی ۲۶ سال اما در ظاهر بسیار جوان تر به نظر می آمد که با شکایت سکسکه های شدید طول کشیده از سه روز پیش و همچنین سردرد با ماهیت فشارنده در ناحیه فرونتال و تهوع و استفراغ ناگهانی جهنده مراجعه نموده بود. بیمار از سکسکه های شدید که حرف زدن را برای وی مختل کرده بود و نیز کاهش شنوائی اخیر و پلی یوری شدید (روزانه ۲۰ بار با حجم وسیع) شاکی بود از دیگر نکات بالینی قابل توجه در بیمار حاضر، وجود ضایعات پوستی نوروفیبروم پلکسی فورم در شش منطقه جداگانه به ابعاد تقریبی ۷ تا ۸ سانتی متر، وجود لکه های شیر قهوه ای از سن سه سالگی و همچنین وجود تومور قلبی به ابعاد ۲×۱ سانتی متر در دهلیز راست و توده ای به ابعاد ۸ × ۷ سانتی متری در لوب آهیانه راست بودند.

بحث و نتیجه گیری: با توجه به شرح حال بیمار و همچنین وجود علائم و عوارض هر دو نوع تیپ نوروفیبروماتوز و نیز سیر پیشرونده بیماری که در نهایت طی مدت چهار روز بعد از تشخیص بیمار منجر به فوت وی شد پاتوژنز سیر برق آسای بیماری و فوت بیمار به روشنی مشخص نشد و تصور می شود که پیرو گزارشات اخیر پیرامون تیپ‌های جدید مورد بحث در خصوص نوروفیبروماتوز این مورد نیز به عنوان تیپ جدیدی از نوروفیبروماتوز مطرح باشد.

واژگان کلیدی: نوروفیبروماتوز، لکه های شیر قهوه ای