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Case Report
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The Co-existence of Celiac Disease and Neurofibromatosis Type 1 in a Child

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Abstract

A 7 year old African-American (AA) boy with Neurofibromatosis type l (NF-l) was seen in the pediatric endocrine clinic for developing breast buds. It was noted that his growth had been decelerating for the previous several years. Investigations for his growth failure included serologic screening for CD. This was abnormal and biopsies of the duodenum were consistent with CD. This is the first pediatric case of the association of NF-l and CD reported in a pediatric patient. Both diseases may cause growth failure. Furthermore, the incidence of CD in AA is very uncommon as compared to Caucasians. CD should be considered in AA with symptoms of CD.

Introduction

Neurofibromatosis type 1 (NF1) is an autosomal dominant genetic disorder with an incidence of 1:3000 [1]. Celiac Disease (CD) is a common disorder and in the developed world effects approximately 1.0% of the population. We report the first case in a pediatric patient with both NF-1 and celiac disease. Our patient was noted to have numerous café-au-lait spots at birth. He was subsequently confirmed to have NF1 as a result of a denovo mutation. His family had immigrated to the USA from Senegal. Neither parent nor other relative had NF.

At age 7 years he was evaluated by pediatric endocrinology for the development of bilateral breast hypertrophy. The physical exam at that time showed a significant deceleration of his weight percentile from the 10th percentile to below 3rd percentile in weight and from the 5th percentile to below 3rd percentile in length (Figure 1). Multiple large Café-au-lait spots as well as subcutaneous nodules on abdomen, chest and extremities were noted. Hypetrophied breast tissue was also noted. Because of the growth deceleration, a work up for sale was pursued. An anti-Tissue Trans-glutaminase (TtG-IgA) was noted to be 81 IU (0 to 20 IU) He was referred to pediatric gastroenterology wherein a repeat TtG-IgA was 65 IU and the anti-Endomysial IgA titer was 1:10. A duodenal biopsy confirmed an abnormality consistent with celiac disease (Marsh lll A). There were no other symptoms of CD. There was no family history of CD. A gluten free diet was instituted.

HLA typing was negative for DQ2 and DQ8. HLA typing revealed DQA1 × 01/ × 03, DQB1 × 0202. The remainder of an extensive work up for growth deceleration was non-contributory. His saliva was sent to a commercial laboratory ("23 and me") which showed his ancestry on the maternal side to be 98% Sub-Saharan.

The patient was recommended to follow gluten free diet and met with a nutritionist well versed in the treatment of celiac disease. On following him over 16 month's period while he has tried to be compliant with gluten free diet. There has been a consistent a downtrend of tTG IgA levels to 26.8 IU. The growth continues to be maintained below 3rd percentile curve for both stature and weight but has not decelerated further.

Discussion

This association of CD with NF1 is the first reported case in the pediatric population of an African-American child. There has been one other report in the recent literature involving a child [2]. There has been one other report of this association in an adult [3].

Growth patterns amongst children with NF1 have been noted to be shifted downward [4]. Although our patient had been at the 5th percentile for most of his life, the deceleration of his growth represents a very subtle change. More importantly, he is African American (AA) and hence CD would be low on the list of conditions that one would think of to account for his growth pattern. Especially given the absence of other symptoms that would suggest CD. However the term African-

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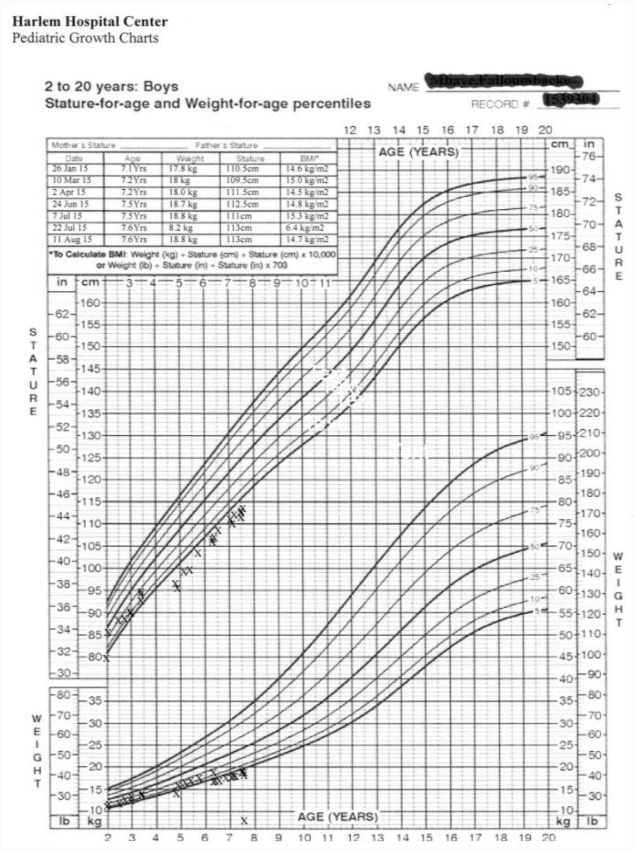

Figure 1: Growth Curve.

American is a relatively recent social construct that encompasses people with significant differences in their genomes and who hail from an enormous part of the world. We do not know the incidence of CD in North America amongst African-Americans but is believed to be quite low [5]. However, in some populations of Africa, like those with Sub-Saharan origins may have an extraordinarily high prevalence of CD [6].

It is also notable is that our patient's HLA typing was negative for both DQ2 and DQ8. The commonly held belief is that this finding would eliminate CD. However there have been studies of patients with CD who are negative for HLA DQ2 and/or DQ8. In some studies, up to 5% of patients with CD other HLA alleles. Our patient had half of the most common HLA allele found in those celiac patients that are both DQ2 and DQ8 negative [7,8].

Summary

We present the first pediatric case of an association between CD and NF1 in an AA male child. This combination of disease is presumably quite rare and may indeed be a random association. On the other hand, we could only find a single reference in which the diagnosis of CD in a population of patients with NFI in a Caucasian child. Hence it is important to remember is that CD should be considered in any population that may have signs or symptoms of CD that have been ascribed to an underlying disease process despite ethnicity of the patient.

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