Case Report

Scurvy in children with autistic spectrum disorder: Not such a rarity

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Abstract

Scurvy is an uncommon nutritional deficiency that results from low serum levels of vitamin C (ascorbic acid). Part of human history since ancient times, its incidence rapidly decreased following the discovery by Sir James Lind that citrus fruits can prevent it[1]. Despite this scurvy still exists today within certain predisposed groups.

We report three cases of scurvy that presented within a short time frame to the Paediatric Department in Mater Dei Hospital (MDH), Malta. All three children were known to have autistic spectrum disorder (ASD) with restricted diets.

A high index of suspicion together with appropriate history and examination can lead to timely diagnosis of this disease. In children deemed at-risk of developing scurvy we recommend routine screening for low serum levels of vitamin C.

Keywords

Vitamin C, Scurvy, Autistic Spectrum Disorder

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Introduction

Scurvy is a nutritional deficiency of vitamin C that is rare in developed countries[2]. Its low incidence and non-specific presenting symptoms often leads to extensive investigations and delayed diagnosis.[1] At-risk groups of children include those with restricted diets stemming from psychiatric or developmental disorders, infants fed evaporated milk, haemodialysis patients and those receiving chemotherapy.[3-4]

Case Presentations

Case 1. A 4 year 11-month-old girl known to have Coeliac disease and autistic spectrum disorder (ASD) presented to the paediatric casualty with recent onset of a limp and left sided hip pain. The limp appeared one week after a viral upper respiratory tract infection; there was no associated trauma. A left-sided antalgic gait with no limb or joint deformity was observed on examination. Over the next few weeks the patient was closely followed up at the child outpatient clinic. On investigation inflammatory and infective arthritides were excluded and iron deficiency anaemia (IDA) was diagnosed. The initial working diagnosis was of a reactive arthritis.

Approximately one month after the initial presentation the patient re-presented to the paediatric casualty with easy bruising over lower limbs, an ulcer over her right big toe, diffuse hair loss, weight loss and a new ulcer over the left upper gum associated with gingivitis/contact bleeding. Further investigations included a bone marrow biopsy, multiple radiological imaging modalities and blood tests. An echocardiogram revealed a 3mm pericardial effusion. The patient continued to deteriorate clinically with a dropping haemoglobin, low grade fevers, persistent easy bruising, increased irritability and refusal to weight-bear having become bed bound.

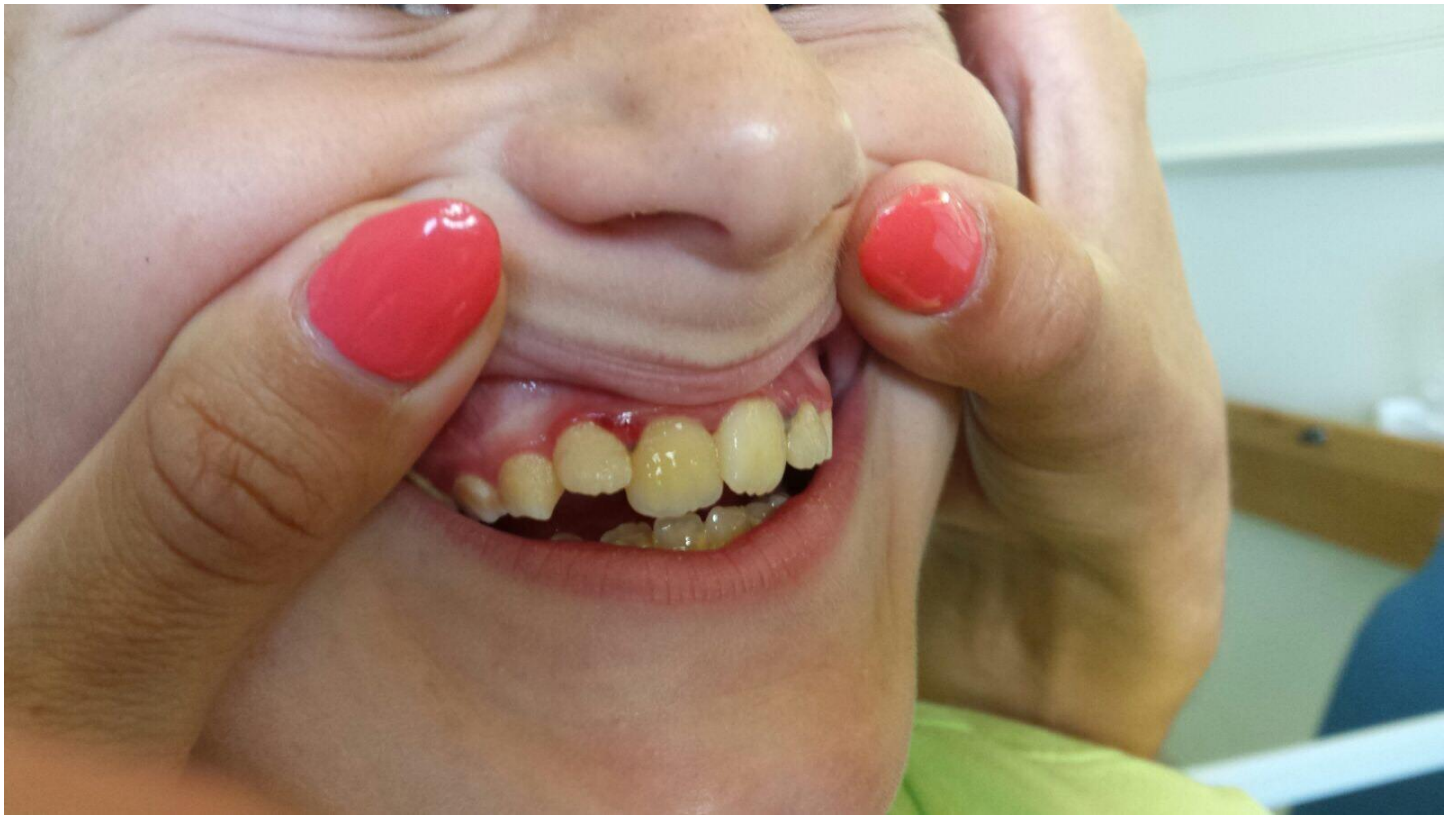
In view of the marked gingivitis, an ulcer of the lower lip, widespread petechial rash, bruising and joint pains, a vitamin C level was assessed

using High Performance Liquid Chromatography (HPLC) and found to be markedly low (<1.0mg/l, normal range 5.0-15.0mg/l). A diagnosis of scurvy was made. Her dietary history revealed years of an extremely restricted diet that had become further limited by her Coeliac disease. A dramatic and rapid improvement in all her symptoms was observed once high dose vitamin C supplementation at a dose of 250mg daily per oral was commenced.

Case 2. A 10-year-old girl, sibling to case 1, also autistic on a restricted diet was asymptomatic. However, routine screening showed a vitamin C (HPLC) level of <1.0mg/l (normal range 5.0-15.0mg/l) confirming the diagnosis of scurvy. She was started off on 20mg vitamin C supplements three times daily by mouth.

Case 3. A 9-year-old boy with ASD presented with an intermittent limp for two weeks. He was able to weight-bear, had no recent illnesses or trauma and on examination had no deformities. His parents described a one-month history of bleeding gums and a recent history of increased lethargy and decreased appetite on a background of a restricted diet. On examination gum hypertrophy and gingivitis (Figure 1) was found. Serum vitamin C (HPLC) levels were found to be <1.0mg/l (normal range 5.0-15.0mg/l). Within weeks of initiating oral replacement therapy, initially with 20mg vitamin C three times daily, his gum hypertrophy and bleeding resolved and his physical activity returned to normal.

Figure 1: *Clinical photograph of Case 3 showing inflamed and hypertrophic gums*



Discussion

Restricted interests and repetitive behaviour are core features of autistic spectrum disorder (ASD) and can result in feeding difficulties, restricted diets and a predilection for nutritional deficiencies including scurvy.[5]

Vitamin C found in citrus fruits and some vegetables (tomato, potato, cabbage, broccoli, lettuce, red peppers)[2] is an essential nutrient which humans are unable to synthesize *de novo*[1] with no stored form. Consequently, even just 1-3 months of an insufficient intake can lead to clinical manifestations of scurvy[4]. Vitamin C plays a major role in the synthesis and cross-linking of intercellular connective tissues, collagen, dentine and osteoid.[2] The resultant increased fragility of these tissues in scurvy explains many of its clinical manifestations.[6] Vitamin C plays other roles in human biochemistry as a cofactor, reducing agent and antioxidant[2] and enhances iron absorption.[1]

The earliest manifestations of scurvy are non-specific constitutional symptoms including lethargy, weight loss, decreased appetite and low-grade fevers.[2] Dermatological manifestations such as petechiae, ecchymoses, perifollicular haemorrhages and corkscrew hairs appear early on in the disease process. Gingival disease usually occurs next with swelling, bleeding, hypertrophy and loosening of teeth or poorly formed teeth.[7] In contrast to adults with the disease, musculoskeletal abnormalities are found in 80% of paediatric patients with scurvy[8] often resulting in an orthopaedic presentation. Joint pain/swelling, myalgias, haemarthrosis, muscular haematomas and fractures can occur resulting in limb pains, limping, leg weakness and/or refusal to weight-bear[6]. Iron deficiency anemia commonly coincides with scurvy secondary to decreased efficacy of iron absorption as well as iron losses from easy bleeding and bruising.[1] Cardiac hypertrophy, pulmonary hypertension and right heart failure have been reported in paediatric groups with scurvy[9] and could explain the persistent small pericardial effusion in Case 1. More advanced scurvy may present with bone marrow and adrenal suppression, psychological changes, poor wound healing and even death.[2]

The diagnosis of scurvy is often elusive, and a high index of suspicion combined with thorough history and examination is pivotal to confirming an early diagnosis without excessive and extensive investigation. Serum vitamin C levels (<2mg/L) are the gold standard for diagnosis, although radiological investigations provide complimentary evidence of the disease.[4] No radiological abnormalities were reported in our cases.

Ultimately, definitive confirmation of scurvy is the rapid resolution of symptoms once treatment with vitamin C supplementation is commenced,[7] with improvement of symptoms starting within 24 hours.[8] No specific treatment regime has been proposed for paediatric scurvy and the oral route is effective even in severe states of deficiency.[4] Weinstein et al. recommend oral doses of 100 to 300mg of vitamin C for paedatric scurvy patients until adequate serum levels are achieved -typically one month after commencing treatment.[8]

Conclusion

Scurvy is a rare disease associated with significant morbidity from the disease process itself and from the excessive sometimes invasive investigations that are undertaken if it is not considered in the differential diagnosis. Heightened awareness of this easily curable disease is especially important in patients suffering from ASD and others with restricted diets. We recommend screening for scurvy in all autistic patients known to have a restricted diet.

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