A 10-year-old boy with Trisomy 21 presented to the emergency department with a history of rash on his legs, easy bruising and bleeding, and walking with a limp. One month prior to presentation, he had developed a non-pruritic, non-painful, petechial rash on his lower legs (Figure 1). Eczema was suspected and he was treated with a topical steroid cream, but there was no improvement in the rash. Subsequently, he developed easy bruising and bleeding gums when brushing his teeth. The day prior to presentation, he began limping and then refused to walk. His parents also reported a several-month history of decreased appetite and poor weight gain. They deny any fever, night sweats, or recent illnesses.

His medical history was also significant for autism, chronic constipation, and very restricted diet for the past 3 years. Over this time, his diet had been limited to peanut butter and jelly sandwiches, soy bacon, pasta, waffles, wheat porridge, cheese crackers, vanilla pudding, toaster pastries, chocolate sandwich cookies, and pretzels. He refused to eat all other foods. He was not taking any supplemental medications, vitamins, or alternative therapies.

**PHYSICAL EXAMINATION**

On presentation, he was noted to be a thin-appearing child who had fine, kinky hair. He was 24.5 kilograms (15th percentile on growth curve for Trisomy 21). He had gingival swelling and bleeding around the base of several teeth, but otherwise good dentition without caries. There was blood in the left external auditory canal obscuring the left tympanic membrane; the right canal and right tympanic membrane were normal. Heart, lung, and abdominal examinations were unremarkable. His left knee was swollen without effusion or tenderness and had full range of motion; the right knee was normal. His skin was remarkable for perifollicular petechiae over bilateral lower extremities, face, and trunk, with scattered bruises over his legs.

**HOSPITAL COURSE**

He was admitted to the hospital for further evaluation. Complete blood count, serum chemistries, liver function panel, and coagulation panel (prothrombin time, activated partial thromboplastin time, and international normalized ratio) were normal. Plain radiographs of the lower extremities were unremarkable. Additional laboratory testing confirmed the diagnosis.
Diagnosis:

Scurvy

The patient was found to have an extremely low level of vitamin C at < 0.12 mg/dL (normal range, 0.2-1.90 mg/dL). Within 3 days of starting vitamin C supplementation, the petechiae, bruising, and gum bleeding had resolved and the patient was walking normally. He was discharged home to continue vitamin supplementation and follow up with his primary care physician.

DISCUSSION

Inadequate intake of vitamin C and resultant scurvy has been described in the elderly, single men, and those who abuse drugs and alcohol. Few pediatric case reports of scurvy exist in the literature, and lack of physician awareness may delay diagnosis. Children with severe eating restrictions may present with multiple nutritional deficiencies or concurrent medical conditions, complicating the detection of scurvy. Hypovitaminosis C may not be revealed until more invasive or expensive testing has been completed, such as bone marrow biopsy looking for underlying malignancy. In this case, the patient initially was admitted to the hematology/oncology service because of concern for leukemia.

Water-soluble vitamin C is a necessary co-factor in various hydroxylation reactions, most importantly those involving lysyl and prolyl residues required for the triple-helix structure of collagen. In an animal model, vitamin C deficiency also reduced the expression of type IV collagen and elastin mRNAs in blood vessels. The abnormal collagen produced in the setting of vitamin C deficiency accounts for the more dramatic symptoms of scurvy (ie, hemorrhage and poor wound healing). Diminished blood vessel integrity leads to bruising, petechiae, edema, and swollen and bleeding gums. Hemorrhage into muscle tissue, joints, or beneath the periosteum causes pain and reduced mobility. Impaired collagen turnover disrupts healing of new wounds, and old scars may thin and reopen.

In addition to collagen formation, vitamin C has many other functions. Kinked or corkscrew hair can result from impaired disulfide bond formation. Anemia, rarely seen in experimental settings, is common in naturally occurring cases, likely due to intestinal bleeding and concomitant deficiencies in folate. Vitamin C has been shown to increase iron absorption two- to six-fold, and to prevent oxidation and excretion of folate. In human experiments, vitamin C deficiency impaired vasoconstriction in response to adrenergic stimuli; this effect may underlie episodes of syncope, ST-segment elevation, and apparent sudden cardiac death. The mechanism by which vitamin C deficiency causes other commonly described symptoms of scurvy, including fatigue, depression, and cardiovascular events, is less understood, but may reflect vitamin C’s involvement in the metabolism of carnitine, corticosteroids, and neurotransmitters.

Humans are dependent on dietary sources of vitamin C, present in abundance in citrus fruits, broccoli, and sweet red peppers, and to some degree in many other fruits and vegetables. Preservation and cooking methods may decrease vitamin C content. At low doses, vitamin C is highly absorbed; higher doses are less avidly absorbed and undergo urinary excretion. Daily intake of as little as 10 mg of vitamin C (present in 20 g of orange or 50 g of potato) has been shown to reverse symptoms of scurvy. Prolonged deficiency, often 3 to 6 months, is required to produce disease.

Children with autism spectrum disorder (ASD) are at greater risk of scurvy and other nutritional deficiencies due to their high likelihood of restricted eating patterns. Narrow food preferences are consistent with other restricted behaviors in children with autism, and food preferences may be complicated by unusual sensory sensitivity to particular textures, smells, or colors. Case reports of other nutritional deficiencies with clinically significant sequelae in children with ASD have been reported, including vitamin B12 and optic neuropathy, vitamin D deficiency, iron deficiency, and anemia. Analyses of detailed food records confirm decreased vitamin C consumption among children with ASD. However, although most children with autism consume a more limited diet with frequent food refusals compared with their typically developing peers, most still meet basic nutritional needs. Because most case reports in the pediatric literature on
scurvy are among children with ASD, caregivers and health care providers should pay careful attention to the severity of eating restrictions in this large and growing population.

Treatment of severe nutritional deficiencies secondary to selective eating patterns in children with autism requires a multidisciplinary team approach with parent-therapist partnership. Experienced therapists may be able to advise families on how to expand nutrient value by utilizing the textures and foods already tolerated by the child (e.g., mixing supplemental vitamins or small amounts of new tastes to favorite foods). To address more restricted preferences, the sequential oral sensory method encourages hierarchical tolerance of food, gradually moving from touching (engaging food in play activity) to ultimately eating foods of different textures, colors, tastes, and smells. This therapy uses similar strategies to those used in other areas of rigidity seen in children with autism, such as expanding types of play and tolerance to daily routines, and should involve the cooperation of behavioral therapists, speech/feeding therapists, and families. Ideally, feeding routines should be included into visual schedules and into concrete school and outside therapy goals.

**CONCLUSION**

Scurvy is a condition resulting from prolonged vitamin C deficiency that can be seen rarely in children with severely restricted diets. Children with ASD, representing an estimated 1% of children in the United States,25 are at greatest risk. Nutritional risk assessment of these vulnerable youth, and referrals to therapeutic specialists when necessary, is an essential part of their holistic medical and developmental assessment.

**REFERENCES**


