

## CASE SERIES

# A spectrum of misdiagnosis to missed diagnosis— A case series of Scurvy in children with intellectual impairment

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### ABSTRACT

Scurvy is caused by prolonged dietary deficiency of vitamin C, an important micronutrient. The varied presentation of scurvy in children makes the diagnosis elusive. We report a case series of scurvy in 7 children with intellectual disability who presented with bleeding and predominant musculoskeletal symptoms. The dietary history revealed a diet deficient in fruits and vegetables. A clinical diagnosis of scurvy was made and that was supported by radiological features in X rays. They were started on oral vitamin C supplementation. All children showed response to therapy in few weeks.

Scurvy is an overlooked diagnosis that is prevalent in certain susceptible group of children with food fads and feeding difficulty.

**Key words:** Scurvy, bleeding, Vitamin C

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### INTRODUCTION

Vitamin C is a water soluble vitamin with a half life of 10 -20 days. Ascorbic acid is a potent antioxidant and cofactor in biosynthesis of collagen, carnitine and neurotransmitters. It is important for the formation of collagen triple helix, conversion of folic acid to folinic acid, synthesis of dopamine, epinephrine, Norepinephrine, carnitine and for metabolism of cyclic nucleotides and prostaglandins. Humans cannot produce vitamin C and are dependent on dietary supply due to mutation in gene coding for L-gluconolactone oxidase required for Vitamin C biosynthesis.

Dietary sources of Vitamin C include fruits like oranges, lemon, melon, kiwi, grapes, strawberries, papaya, gooseberries and guava and vegetable sources include cabbage, asparagus, tomatoes, sprouts, spinach, bell peppers and broccoli. Foods lose Vitamin C on prolonged cooking and there is no storage of Vitamin C in the body.

The RDA of Vitamin C in children are 50 mg for infants 6-12 months, 15 mg for children 1-3 years, 25 mg for 4-8 years and 45 mg for 9-13 years.

Dietary deficiency of Vitamin C for 8-12 weeks causes clinical symptoms of Scurvy. Vitamin C depletion causes structural alterations in collagen, defective osteoid matrix and dentin formation and increased bone resorption. Capillary fragility leads to bleeding tendencies.

The causes for scurvy include selective unbalanced diet, food behavioral disorders, Neurological disorders (cerebral palsy, mental retardation, Autism spectrum disorder), children on chemotherapy, post bonemarrow transplant. Scurvy in paediatric age group is underdiagnosed and scantily reported. The wide spectrum of clinical presentation results in misdiagnoses of Scurvy as septic arthritis, osteomyelitis, Leukaemia, bleeding disorders, child abuse and juvenile idiopathic arthritis and children with scurvy present to different specialities like Orthopedics, Paediatrics, Dermatology, Dentistry, Rheumatology, Oncology, Neurology and Ophthalmology.

**CASE SERIES****CASE 1:**

6 years old female child a case of cerebral palsy with mental retardation was admitted to orthopedics department as she presented with fever, joint pain and swelling both knees with restricted mobility.(Figure 1) Septic arthritis was suspected and child was started on antibiotics and diagnostic arthrocentesis was done. As arthrocentesis showed blood, child was referred to Paediatrics for evaluation. Detailed history taking revealed gum bleeding. Complete hemogram showed Hemoglobin 8.2 gm, Total count 12000 and platelet count 180000 and normal coagulation profile. X ray of the joint showed pencil thin cortex and subperiosteal haemorrhages.



**Figure 1: Bilateral knee joint swelling with skin bleeds**

**CASE 2:**

5 years old female child a case of Mental retardation brought with history of sudden protrusion of left eye with pain and diminished vision. Child was examined at Ophthalmology OPD and referred to Paediatrics for further evaluation. History revealed gum bleeds and joint pain involving both knee joints in the past. Clinical examination showed Proptosis of left eye and swelling of both knee joints. Complete hemogram showed anemia and coagulation profile was normal. CT of the left orbit showed retrobulbar haemorrhage. Xray of the lower limbs with knee joint showed osteopenia, pencil thin cortex and pelkan spur.

**CASE 3:**

10 years obese male child with mental retardation was brought with history of suspected trauma and difficulty in walking to trauma casualty. Past history revealed previous similar episodes. Clinical examination showed swollen ankle joints and perifollicular haemorrhages and pseudoparalysis. A clinical diagnosis of scurvy was made.

**CASE 4:**

8 years old female child, a case of mental retardation brought with fever, bleeding gums and pain both knee joints. Clinical examination of the child showed pallor, swollen gums with swelling both knees and painful restriction of movements of both knees. Lab investigations were normal but for iron deficiency state. X ray of the knee showed osteopenia with ground glass appearance.

**CASE 5:**

A 6 year old female child a known case of Autism, with history of multiple visits to dentists for gum bleeding and loose teeth was brought for evaluation. Clinical examination showed follicular haemorrhages with hyperkeratosis and swollen gums. Coagulation profile was normal and Xray showed osteopenia, pencil cortex and Wimberger ring sign.(Figure 2)



**Figure 2: X ray knees showing Trummerfeld zone, pencil thin cortex, Wimberger sign and Pelkan spur**

**CASE 6:**

A 12 year old male child a case of Leukodystrophy was admitted with fever, irritability and joint swelling involving both knees. Clinical exam showed anaemia, contractures with swollen knee joints. Xray revealed ground glass appearance and fracture of lower end of femur with epiphyseal separation.

**CASE 7:**

A 3 year old male child with spastic cerebral palsy was brought with history of swollen knee joints and pain on handling. Clinical examination showed swollen bleeding gums, Bilateral knee joint swelling and pseudoparalysis. Xray of the knee joint showed wimberger sign and huge subperiosteal haemorrhage. (Figure 3)



**Figure 3: X ray knees showing subperiosteal haemorrhage**

The most common presenting complaint was arthralgia, involvement of bilateral knee joints predominantly and weakness. A detailed dietary history showed that all children were predominantly

on diet devoid of fruits and vegetables and the reason being feeding difficulty and lack of dietary awareness among care givers .All 7 children showed malnutrition with iron deficiency anaemia.

Serum vitamin C levels were not done due to non availability of the test in the hospital and financial constraints. Based on the clinical features, supportive history and radiological features a diagnosis of Scurvy was made and all children were started on Oral Vitamin C 300 mg/day which was continued for 4- 6 weeks. Analgesics were also given. A suitable dietary modification was done and oral iron was also started. All children responded to therapy with lessened irritability and cessation of bleeding within a week.

## DISCUSSION

Scurvy is seen in children with intellectual disability as they are predominantly fed on milk based diets deficient in fresh fruits and vegetables due to feeding difficulty ,oromotor dysfunction, pseudobulbar palsy ,drooling and difficulty in swallowing solids.Such children show malnutrition with iron deficiency anaemia and multiple vitamin deficiencies.

Vitamin C deficiency presents with non specific systemic symptoms like irritability, malaise, fever, myalgia, loss of appetite and crying when handled. Other symptoms include Dermatological: petechiae, ecchymosis, follicular hyperkeratosis, perifollicular haemorrhages, cork screw hairs Dental: swollen gums, haemorrhagic gingivitis, loosening of teeth Musculoskeletal symptoms(80%): Joint swelling, arthralgia, Limping, Fractures, refusal to walk, pseudoparalysis, scorbutic rosary, muscular hematomas Haematological: Anaemia (secondary to bleeding,decreased iron absorption and abnormal folate metabolism) Ophthalmologic:Orbital haemorrhage, proptosis Others: Intracranial bleed, hemopericardium, haemorrhagic pleural effusion, Pulmonary hypertension, alopecia, delayed wound healing Diagnosis of scurvy is made by a combination of positive clinical findings,radiological findings and response to therapy.Measurement of Serum Vitamin C levels is specific but not sensitive and values less than 0.2mg/dl is considered as deficiency. The vitamin C level in buffy coat of leucocytes correlates well with tissue stores and levels <7 mg/dl is considered as deficiency.Urinary excretion of vitamin C after oral or intravenous supplementation can also be used to diagnose scurvy. These lab investigations are costly and not available in many centers.

Radiological features of scurvy include osteopenia, fractures, pencil outlining of epiphysis and diaphysis, thin bone cortex, white line of Frenkel, Trummerfeld zone, pelkan spurs, wimberger ring sign, scorbutic zone, subperiosteal haemorrhages and epiphyseal separation.

Response to supplementation begins within 24 hours with decrease in irritability and anorexia and pain decreases in 2-4 days, gingival lesions heal in 1 to 2 weeks. Bone abnormalities will resolve over several weeks and a complete resolution of findings occurs by 3 months.

## CONCLUSION

Scurvy though uncommon is still prevalent in certain susceptible children.A high index of suspicion with detailed dietary history and good clinical correlation will help in diagnosing scurvy and avoid unnecessary battery of investigations, delay in diagnosis, shunting between specialists and prolonged antibiotic course.

## REFERENCES

1. A case of Scurvy in a child:An uncommon but important diagnosis to consider- Aziza Elouali, Zohair El Haddar, YasseBouabdella,MariaRkain,AbdeladimBabakhouya, Cureus May 2023;15(5):e39369
2. An unusual presentation of Cerebral Palsy- Sumit Gupta, Rajesh Kanojia, Ashish Jaiman, Dhananjaya Sabat, World Journal of Orthopedics May 2012,3(5):58-61
3. Highly selective eating in Autism spectrum disorder leading to scurvy- A series of 3 patients – Rana Swed-Toli,A.Haj,Aviuel YB,D Militianu,Ram Weiss Paediatric Neurology Volume 94,May 2019,P 61-63
4. Lessons learned from “the great mimicker disease” :A retrospective study of 18 patients with Scurvy- Faisal Miraj,I Wayan Arya,MahendraKarda,Eugene Dionysios, Journal of Children’s Orthopedics 2023 Dec;17(6): 618-625
5. PaediatricScurvy:when contemporary eating habits bring back the past- Alice Brambilla,C.Pizza,D.Lasagni,L.Lachina, RestiM,Trapani S- Frontiers in Paediatrics May 2018,Vol 6
6. Scurvy in paediatric age group- A disease often forgotten? Anil Agarwal,AbbasShaharyar,AnubratKumar,Mohd Shafi Bhat,Madhusudan Mishra; Journal Clinical Orthopedics and Trauma 2015Jun, Vol 6 ,p 101-107
7. Scurvy in children Down but Not Out- Kaur Sukhjot, Goraya Jatinder Singh, Indian Journal Of Paediatric Dermatology Apr- Jun 2021,22(2):p 118-122.