

Nutritional Diseases

OPTH 727



Disease/dysfunction may result from too much or too little of a particular nutrient. (nutrients are fats, carbs, protein, vitamins/minerals)

- What causes someone to have too little of a particular nutrient?
 - Nutrient not ingested
 - Nutrient is ingested, but not absorbed by the digestive tract into the bloodstream (malabsorption)

OR

- Nutrient may be ingested and absorbed into blood stream- but not adequately/efficiently metabolized/stored. A few examples:
 - Acquired metabolic diseases – for example, diabetes mellitus
 - Organ failure: e.g. liver dysfunction
 - Liver has many key functions... several are related to metabolism and storage of nutrients. A few of them are...
 - Glycogen storage and glucose mobilization
 - Fat metabolism and storage
 - Storage of fat soluble vitamins)
 - Inherited metabolic diseases – Gaucher disease, Hunter syndrome, Niemann-Pick disease, PKU, Tay-Sachs, Wilson's disease, hemochromatosis



What are some reasons why someone may not ingest enough of a necessary nutrient:

- Lack of knowledge
 - Patient does not know what foods constitute a healthy diet
- Poverty
 - Insufficient funds to purchase healthy foods
- Unavailability of foods
 - Examples: (famines, crop failure).
 - Crop infection caused potato famine in Ireland in 1840's
 - Lack of fresh fruit and vegetables onboard ships during age of exploration
 - Lack of Vitamin C → Scurvy
- “Fad” diets
- Neglect
 - Persons who cannot care for themselves (children/elderly/disabled)
 - Persons who can care for themselves but chose not to (personal neglect)
- Psychological diseases
 - (anorexia/bulimia)
 - Developmentally disabled children (e.g. autistic)



What are some reasons why a nutrient may not be absorbed into the bloodstream (malabsorption syndromes)?

- There are 4 main elements to absorption of nutrients
 - Pancreas secretes digestive enzymes into the gut- necessary for breakdown of macromolecules (fats, carbs, proteins) that have been ingested
 - Pancreatic diseases will reduce ability to secrete enzymes
 - Pancreatic diseases have many causes but one in particular is alcoholism
 - Liver secretes bile acids needed for solubilization and absorption of fats
 - Liver disease will impede this process
 - Intestinal mucosa is specialized for absorption: mucosal folds and villi produce large surface area
 - Enzymes at intestinal brush border hydrolyze large molecules

Therefore, disruption to any of these processes may result in the inability to absorb nutrients. Diseases of pancreas or liver; status post surgery, parasites, inflammatory diseases, or structural changes to gut may produce malabsorption.

Also blockage of bile ducts (gallstones) will impede bile secretion to gut and possibly also pancreatic enzyme secretion into the gut.



Disease may result from too much or too little macronutrients (protein, carbs, fat).

- Too much
 - Short term excess:
 - Indigestion, GI symptoms,
 - Long term excess:
 - Obesity, cardiovascular disease, diabetes,
- Too little
 - Short term:
 - Fatigue (too few calories consumed)
 - Long term:
 - Muscle wasting and atrophy.
 - Dysfunction of body systems
 - Examples: Immune compromise, brain/kidney/liver dysfunction
 - Long-term protein deficiency
 - Leads to edema (fluid build-up in the tissues)
 - Why? Albumin is a blood protein that is essential for correct osmotic balance (keeps fluid in blood vessels)
 - Lack of albumin - fluid will escape blood vessels and spill into surrounding tissues; causing edema.



What diseases result from too much or too little micronutrients (vitamins and minerals)

- Most diseases result from micronutrient deficiency not excess.
 - Diseases that result from vitamin and mineral deficiency (see next slide)
- Diseases that result from micronutrient excess:
 - Excess iron (iron overload)... called hemochromatosis
 - Iron is a critical component of hemoglobin.
 - Lack of iron produces anemia
 - What is anemia? A deficiency of red blood cells or hemoglobin resulting in weakness and fatigue
 - Iron levels are a tightly controlled process in the GI tract.
 - Hemochromatosis is NOT usually caused by excess dietary iron
 - It is caused by inherited genetic mutation of genes related to iron absorption and storage
 - Or too much iron from frequent blood transfusions
 - Excess iron causes damage to heart, liver and endocrine glands
 - Excess copper:
 - Wilson's disease results from inherited genetic mutation of gene related to copper storage
 - Excess copper is toxic to brain and liver (and other organs)
 - One unique sign of Wilson's disease is brown ring around iris



Please know this slide very well

Vitamin/Mineral deficiencies

Nutrient	also known as	Deficiency causes
Vitamin A	Retinol	Varying levels of vision impairment
Vitamin B1	Thiamin	Beriberi: cardiovascular and neuronal impairment
Vitamin B2	Riboflavin	
Vitamin B3	Niacin	Pellagra: dermatitis (roughened skin), dementia, diarrhea
Vitamin B6	Pyridoxine	
Vitamin B12	Cobalamin	Pernicious anemia
Vitamin C	Ascorbic Acid	Scurvy (see subsequent slides)
Vitamin D		Rickets (children) Osteomalacia (adults) Poor mineralization of skeleton - bone fragility and weakness
Vitamin E	α -tocopherol	
Vitamin K		Inadequate blood clotting
Iron		Iron deficiency anemia



Pellagra - Niacin (Vit B3) deficiency

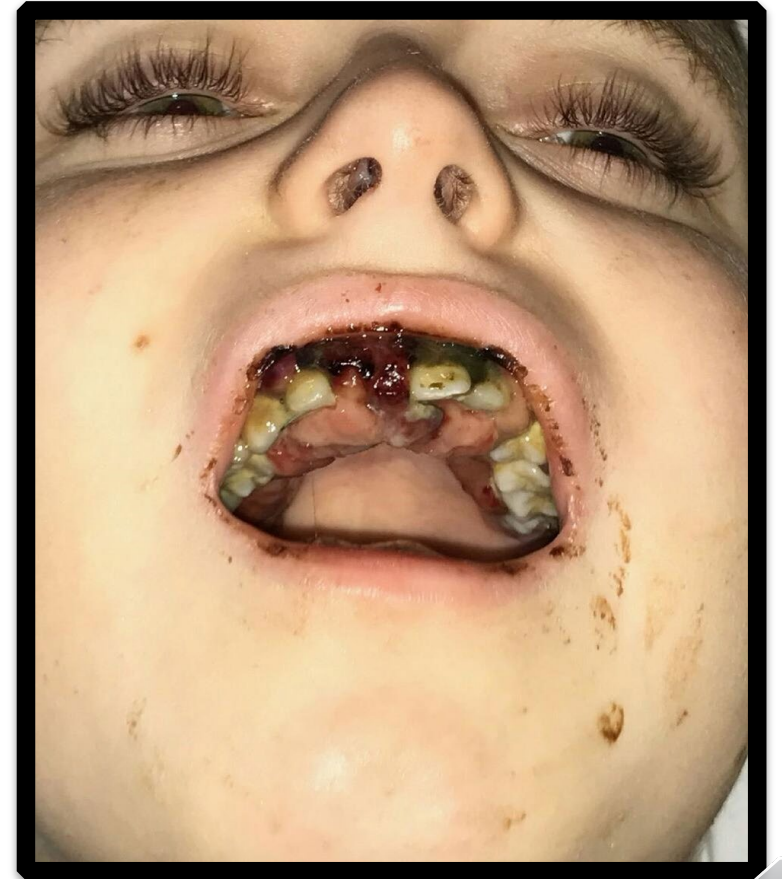


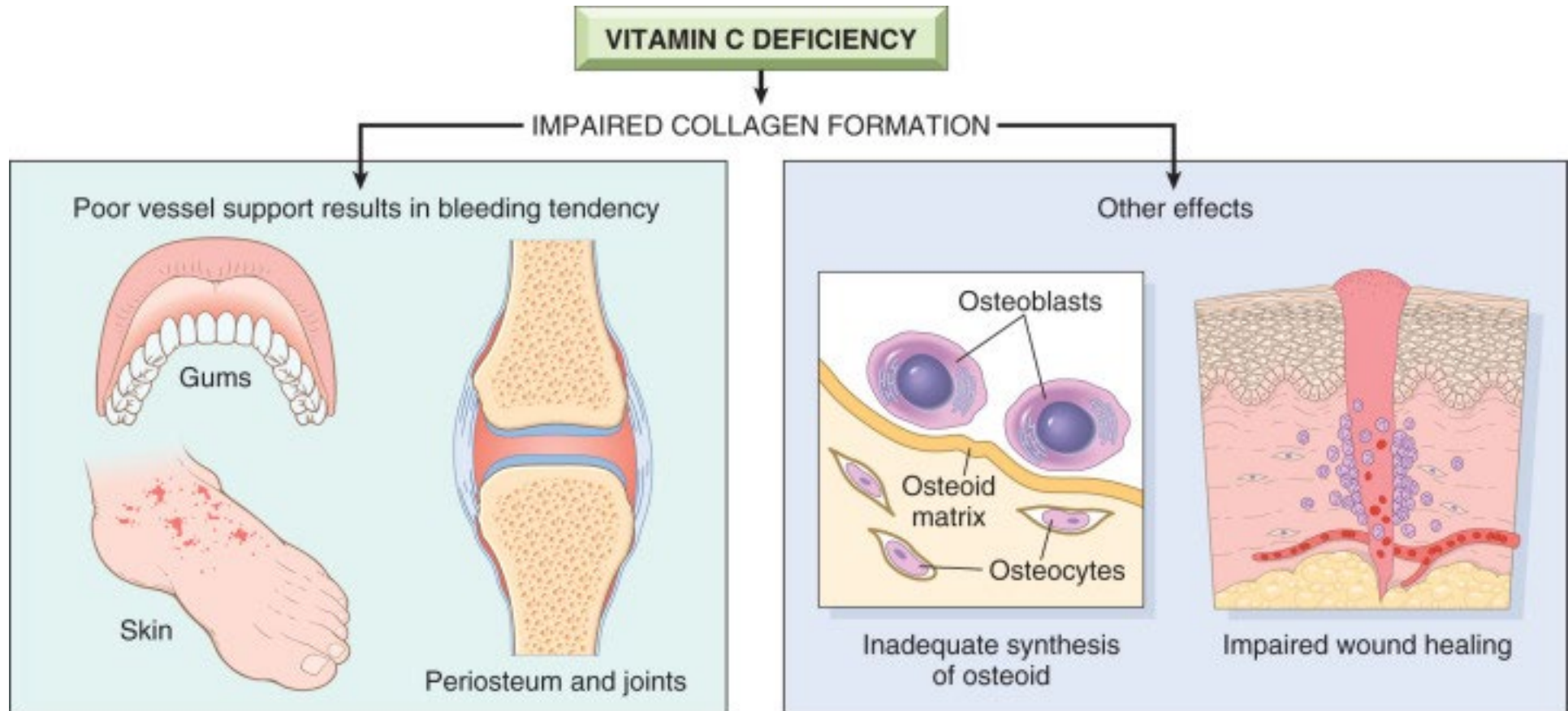
Roughened skin (a type of dermatitis) is one symptom;

The three D's: dermatitis, dementia, Diarrhea



Vitamin C deficiency - scurvy

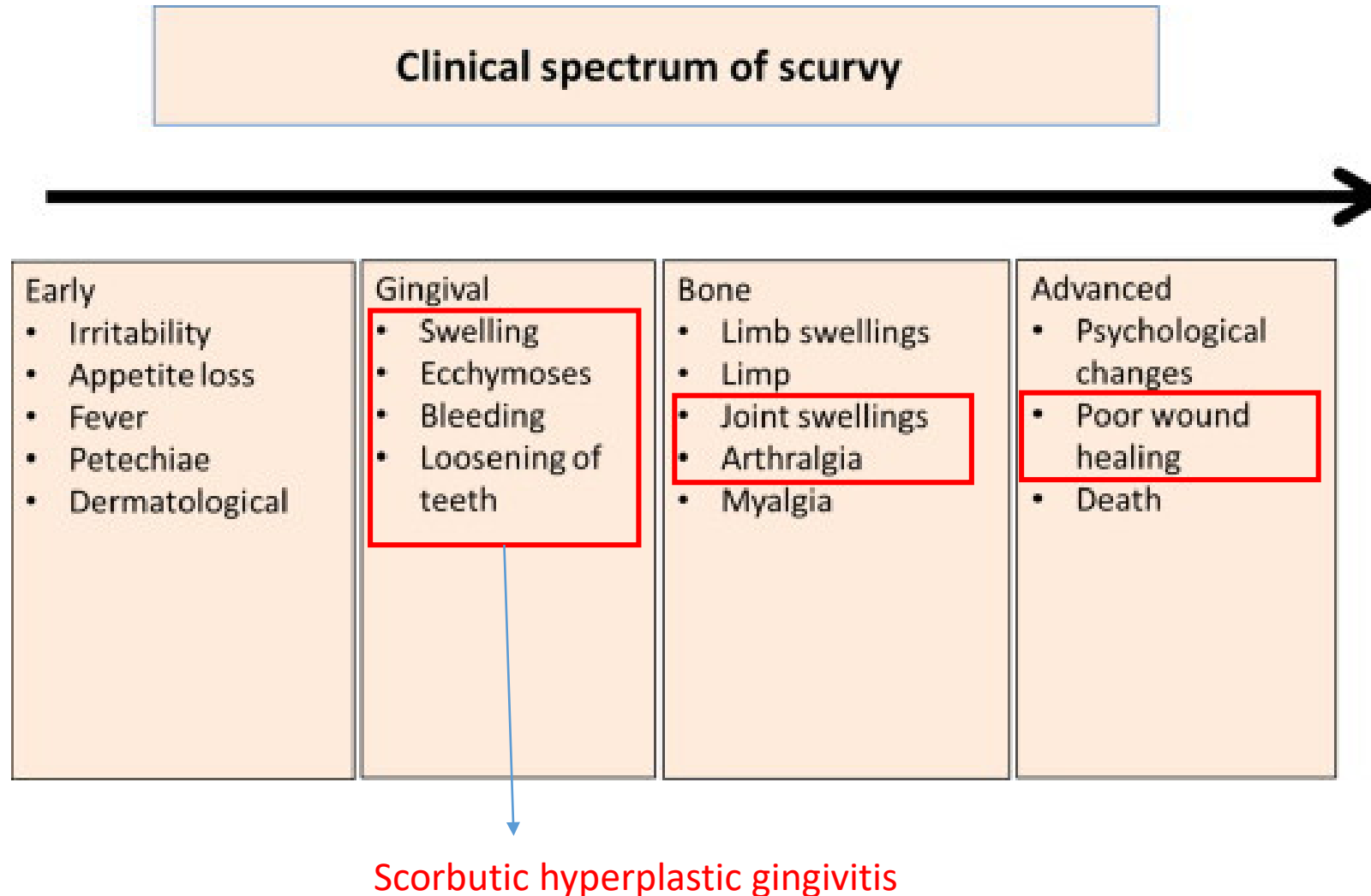




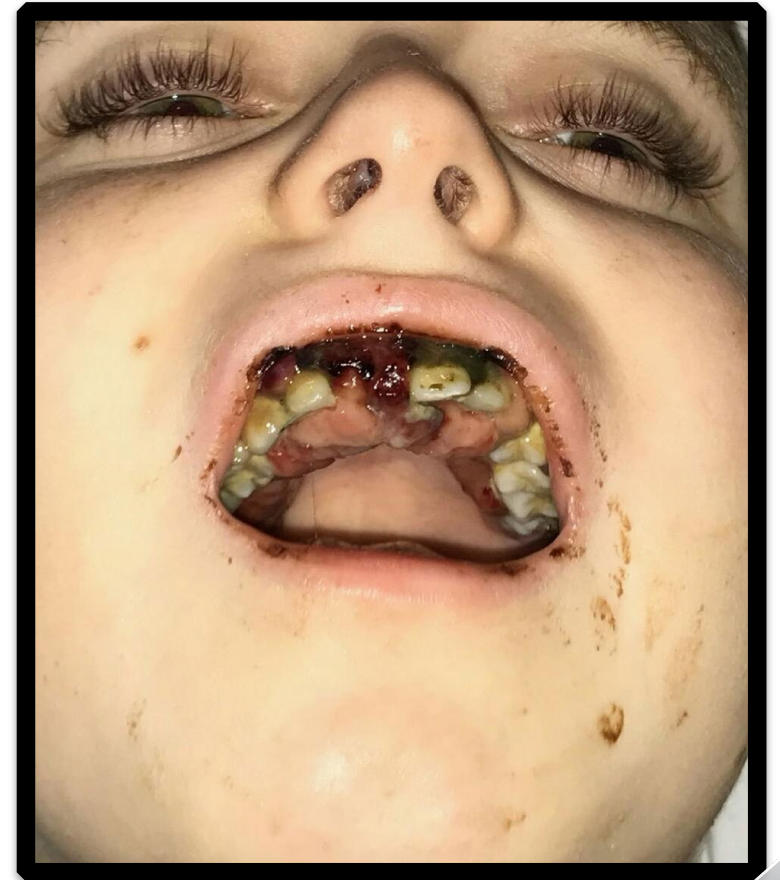
Bleeding into joints results in joint pain and swelling



The symptoms in the red box are most important

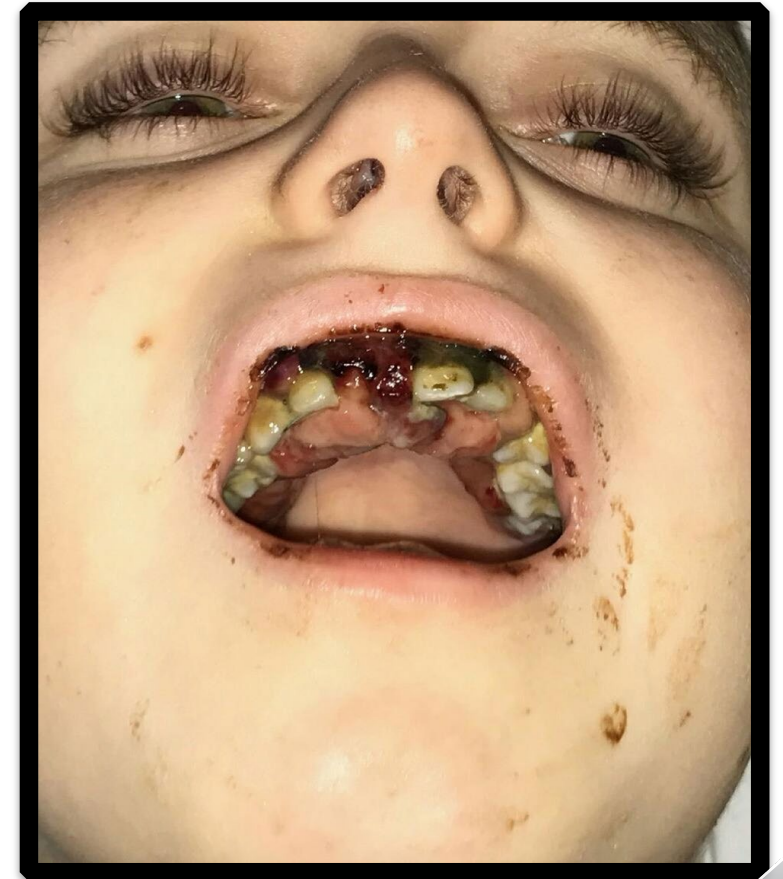


Case of scurvy



Initial Presentation

- 11 year old male
- Pediatric Emergency Department:
 - Severe anemia
 - Bilateral knee pain
 - Spontaneous gingival hemorrhage



Review of Medical History

- Autism, anxiety
- Medications: Prozac
- Diet: pancakes, chicken fingers - exclusively
- Non-ambulatory due to joint pain and muscle weakness

Additional findings:

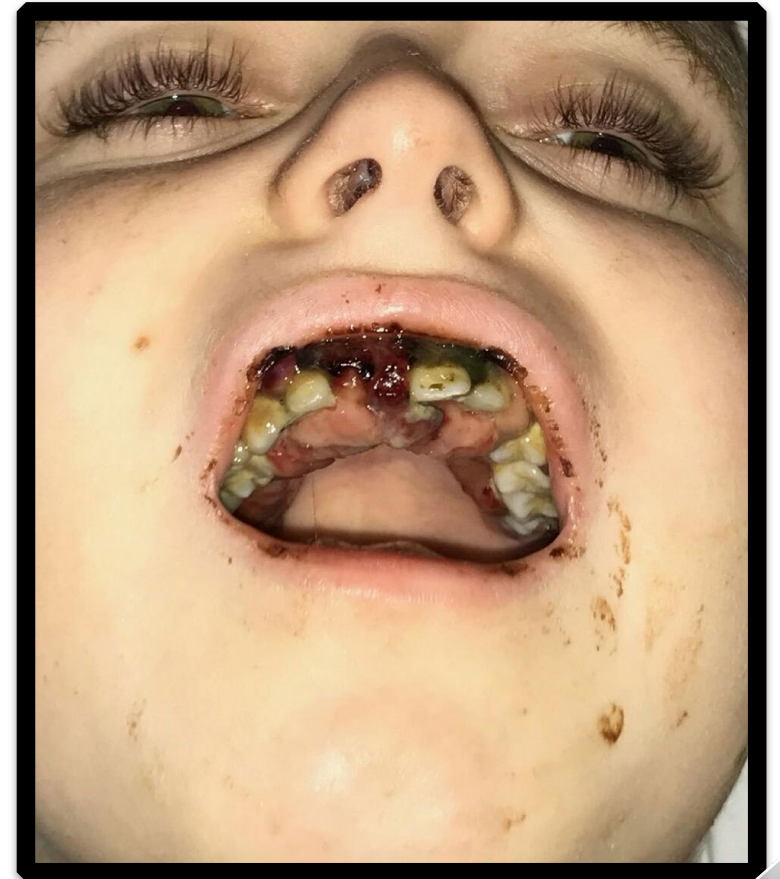
- Rash on the lower extremities,
concern for Henoch-Schonlein purpura

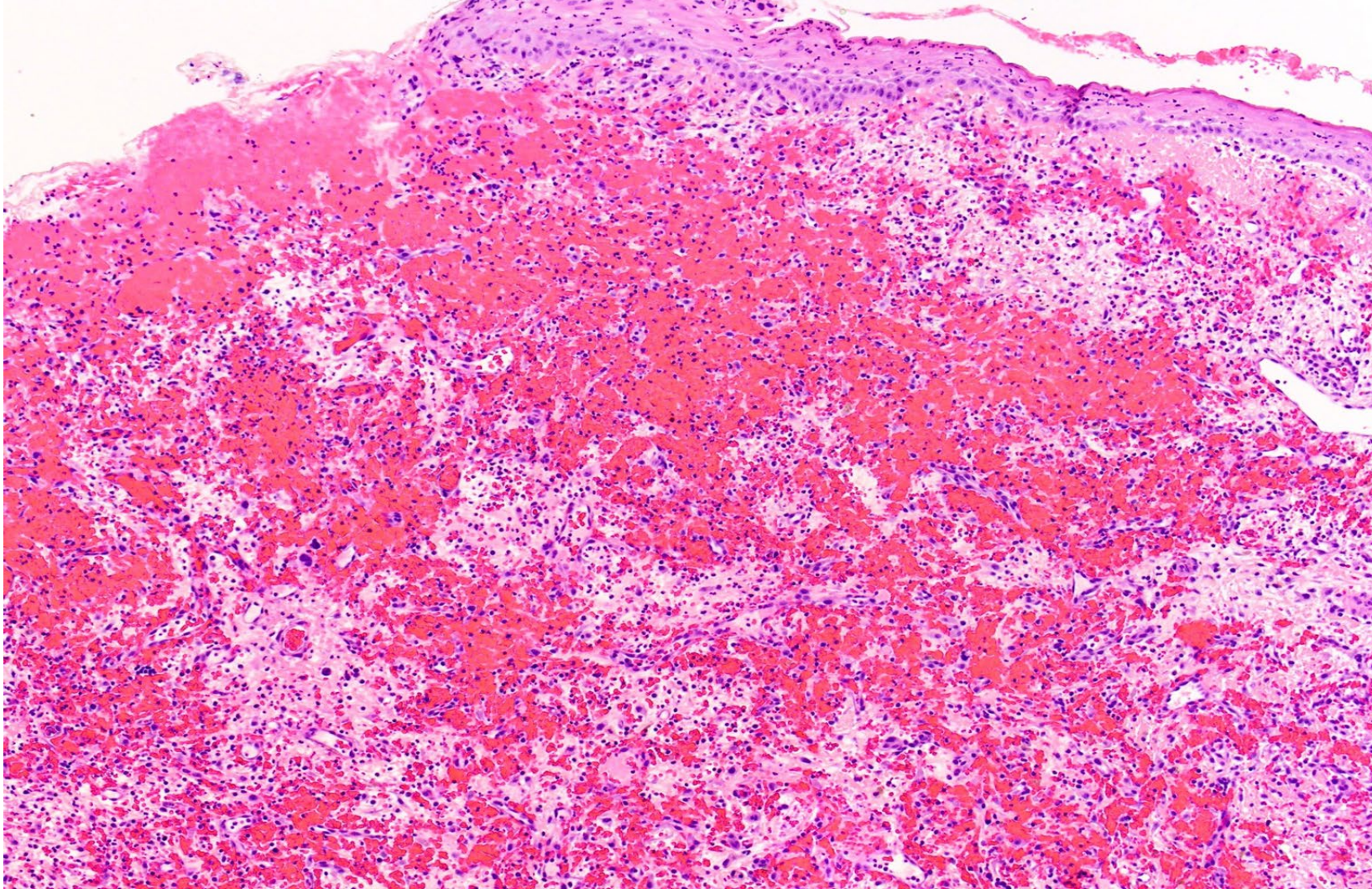


So, what did we do?

We were suspicious of scurvy...

We had to biopsy the tissue. Why did we biopsy? Had to rule out very serious diseases like leukemia.





What did biopsy show?

- 1) Hemorrhage and lack of cohesive connective tissue... suggestive of scurvy.
- 2) Lack of neoplasia.... No leukemia or other cancerous condition.



Bloodwork- Vitamin C levels

Miscellaneous - Chem

Vit C	0.0 L	Ref Range 0.2-2.0 mg/dL
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Patient also had low iron levels.



Treatment Protocol

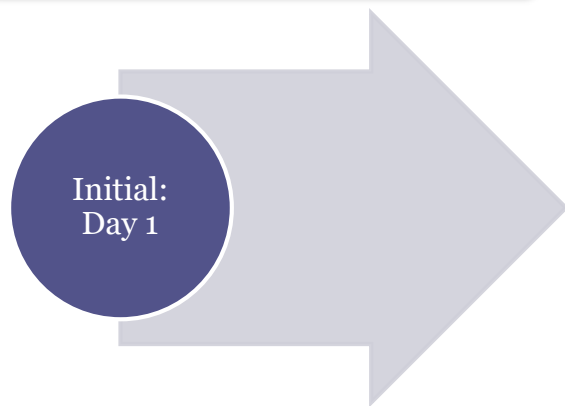
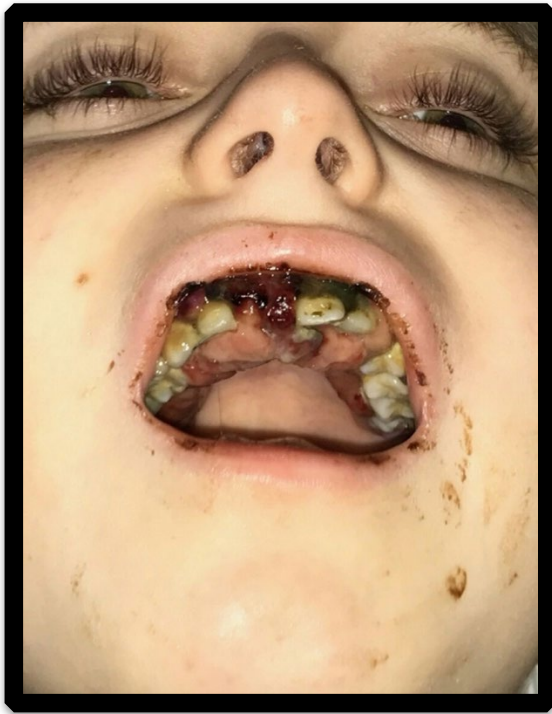
Vitamin C Supplementation

- Children: 100-300 mg/day
- Adults: 500-1000 mg/day

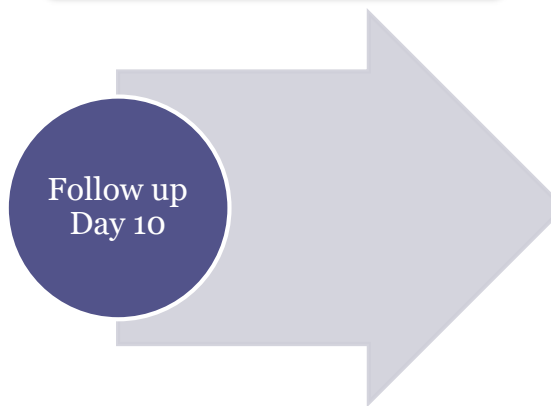
For one month or until symptoms resolve

Additional iron and vitamin supplements.

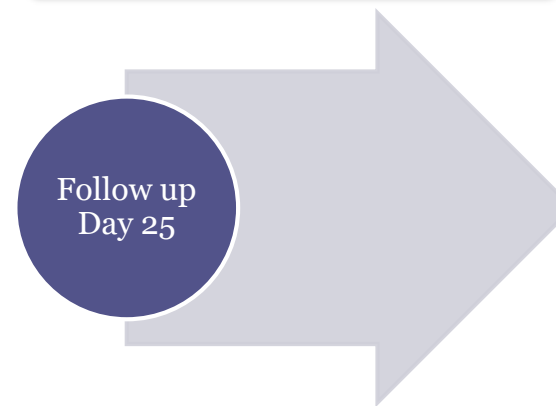
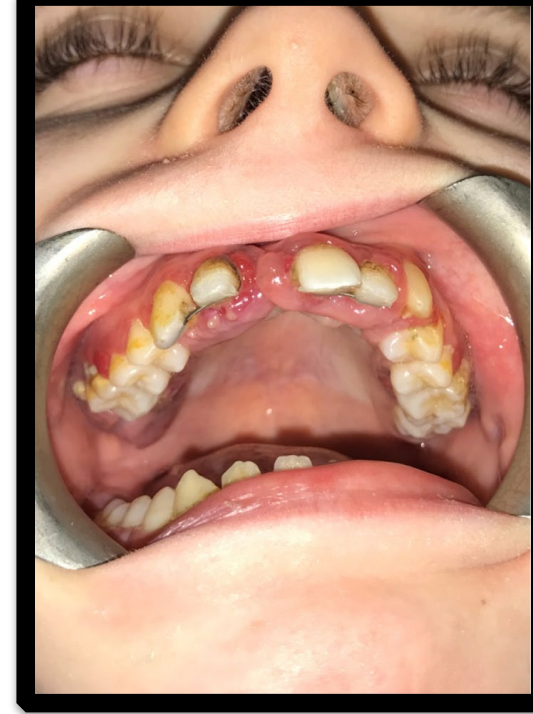




Initial:
Day 1



Follow up
Day 10



Follow up
Day 25

