**CLINICAL CASES FOR THE 2nd YEAR**

**Prepared by**

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**CASE (1)**

**A young man** entered the physician office complaining of **intestinal gases and diarrhea**. He had **signs of dehydration** . The patient **temperature was normal**. He said that this occurred **after** a birthday party in which he had participated in **icecream eating contest .** The patient reported **prior episodes of similar nature following ingestion of a significant amount of dairy products** .

**The clinical picture is probably due to deficiency in :**

A - Salivary α- amylase.

B - Isomaltase.

C - Pancreatic α- amylase.

D - Sucrase.

E – Lactase.

**ANSWER : E – Lactase**.

**LACTOSE INTOLERANCE**

**DEFINITION:**

The inability to digest lactose (the main sugar in milk) due to deficiency of the intestinal enzyme lactase giving rise to gastrointestinal symptoms.

**Lactase deficiency is not the same as lactose intolerance.**

Persons with milder deficiencies of lactase often have no symptoms after the ingestion of milk.-

- For unclear reasons, even persons with moderate deficiencies of lactase may not have symptoms.

- A diagnosis of lactase deficiency is made when the amount of lactase in the intestine is reduced, but a diagnosis of lactose intolerance is made only when the reduced amount of lactase causes symptoms.

**CAUSES:**

**1-Congenital lactase deficiency:**

because of a congenital absence or deficiency (absent from birth) of lactase due to a mutation in the gene that is responsible for producing lactase. This is a very rare cause of lactase deficiency, and the symptoms of this type of lactase deficiency begin shortly after birth.

**2-Primary lactose intolerance**

-- It is the most common cause of lactase deficiency

-- The age of onset and prevalence of this type of lactase deficiency in different ethnic groups is highly variable. Approximately 20% of Hispanic, Asian,and black children younger than 5 years of age have evidenceof lactase deficiency , whereas whitechildren typically do not develop symptoms of lactose intoleranceuntil after 4 or 5 years of age. The clinical importance of these observationsis that children with clinical signs of lactose intoleranceat an earlier age than is typical for a specific ethnic groupmay need an evaluation for an underlying cause, because primarylactase deficiency would otherwise be unusual at such a youngage.

-- Lactose intolerance attributable toprimary lactase deficiencyis uncommon before 2 to 3 years ofage in all populations; whenlactose malabsorption becomes apparentbefore 2 to 3 years ofage, other causes must be sought.

-- Although primary lactase deficiency may present with arelatively acute onset of milk intolerance, its onset typicallyis subtle and progressive over many years. Most lactase-deficientindividuals experience onset of symptoms in late adolescenceand adulthood.

**3-Secondary lactose intolerance**

This type of deficiency is due to diseases that destroy the lining of the small intestine along with the lactase. Causes include:

1- acute infection (eg,rotavirus) causing small intestinal injury with loss of thelactase-containing epithelial cells from the tips of the villi.The immature epithelial cells that replace these are often lactasedeficient, leading to secondary lactose deficiency.

2- Giardiasis and other parasitesthat infect the proximal small intestine often lead to lactosemalabsorption from direct injury to the epithelial cells bythe parasite.

3-some disease of the small intestine as immune-related and other enteropathies

4- Young infants with severe malnutrition develop small intestinalatrophy that also leads to secondary lactase deficiency

**BIOCHEMICAL BACKGROUND:**

**• Normally:**

Disaccharides cannot be absorbed through the wall of the small intestine into the bloodstream

**Small intestinal lactase**

**Lactose Glucose + Galactose Absorbed**

**In lactose intolerance: •**

**In small intestine**

**Small intestinal lactase**

**Lactose Glucose + Galactose**

**Colon**

**In colon**

**Colonic bacteria secreting lactase**

**Lactose Glucose + Galactose**

**Used and some splitted by these bacteria (fermentation)**

**CO2 gas+ Hydrogen gas + acids as lactic acid→acidic stool**

changed into methane gas by another type of colonic bacterium present in some people. These people will excrete only methane or both hydrogen and methane gas in their breath and flatus.

Some is absorbed from the colon and into the body

expelled by the lungs in the breath

A small proportion

is expelled

increased flatulence

(passing gas)

Most of the hydrogen is used up in the colon by other bacteria

-- The copious amounts of gas (a mixture of hydrogen, carbon dioxide, and methane), may cause a range of abdominal symptoms, including abdominal distension, flatulence, acid reflux ,nausea . When sufficient intestinalgas is produced by the bacterial metabolic processes to causestimulation of the intestinal nervous system by intestinal distention,visceral (abdominal) cramping results.

-- Not all of the lactose that reaches the colon is split and used by colonic bacteria. The unsplit lactose in the colon and its fermentation products draws water into the colon (by osmosis). This leads to loose, diarrheal stools.

**The severity of the symptoms of lactose intolerance** varies greatly from person to person as the severity depends on:

*1- The amounts of lactose in diet;* the more lactose in the diet, the more likely and severe the symptoms.

*2- The severity of lactase deficiency,* that is, they may have mild, moderate, or severe reduction in the amounts of lactase in their intestines. Thus, small amounts of lactose will cause major symptoms in severely lactase deficient people but only mild or no symptoms in mildly lactase deficient people.

*3- The different responses of people to the same amount of lactose reaching the colon.* Whereas some may have mild or no symptoms, others may have moderate symptoms. The reason for this is not clear but may relate to differences in their intestinal bacteria.

**INVESTIGATIONS:**

**1-Elimination diet test:**

-- A diet that eliminates milk and milk products, continued long enough to clearly evaluate whether or not symptoms are better

--Elimination of all milk products should eliminate symptoms completely if lactose intolerance alone is the cause of the symptoms

**2-Milk challenge test;**

A person fasts overnight and then drinks a glass of fat free milk in the morning. Nothing further is eaten or drunk for 3-5 hours. If a person is lactose intolerant, the milk should produce symptoms within several hours of ingestion. If there are no symptoms or symptoms are substantially milder than the usual symptoms, it is unlikely that lactose intolerance is the cause of the symptoms.

Milk used must be fat-free to eliminate the possibility that fat in the milk is the cause of symptoms.

If allergy to milk is suspected that primarily occurs in infants and young children pure lactose can be used instead of milk).

**3-Breath test:**

The hydrogen breath test is the most convenient and reliable test for lactase deficiency and lactose intolerance. For the breath test, specific amount of pure lactose is ingested with water after an overnight fast.Samples of breath are collected every 10 or 15 minutes for 3-5 hours after ingestion of the lactose, and the samples are analyzed for hydrogen and/or methane.

If hydrogen and/or methane are found in the breath, it means that the person is lactase deficient.

**4- stool acidity test**

Is a test for lactase deficiency in infants and young children. The infant or child is given a small amount of lactose orally. A lactase deficient infant or child will develop an acidic stool .

The stool acidity test is not done frequently due to superiority of breath testing has led to modifications in the equipment for collecting breath samples that makes it easier to do breath test.

**TREATMENT: According to the severity and age;**

-- Reduction of dairy products in diet to the level it cause no symptoms.

--Small amounts of lactose spaced throughout the day and consumed with otherfoods may be tolerated with no symptoms.Ingestion of other solid foods delays gastric emptying,providing additional time for endogenous lactase to digest dietarylactose.

- Use of partially digestedproducts (such as yogurts,cheeses, andpretreated milks with lactase). Aged cheeses tend to have lower lactose content thanother cheeses and, thus, may also be better tolerated. In case of yogurt . bacteria used to make it contain lactase, and the lactase is able to split some of the lactose during storage of the yoghurt as well as after the yogurt is eaten (in the stomach and intestine). Yoghurt also has been shown to empty more slowly from the stomach than an equivalent amount of milk. This allows more time for intestinal lactase to split the lactose in yoghurt, and, at least theoretically, would result in less lactose reaching the colon.

--Substitutes for cow milk based on rice, soy,or other proteins are readily available and are generally freeof lactose, although the nutrient content of most of these milksis not equivalent to cow milk.

--Oral lactase-replacement capsules(These capsules or predigested milk or dairyproducts with lactase are readily available and will often permita lactose-intolerant individual to be able to take some or allmilk products freely.

--Lactose free milk formula for infants.

--Ca and Vitamin D supplements maybe needed.

**For individuals who are intolerant to even small amounts of lactose, the dietary restrictions become more severe. Any purchased product containing milk must be avoided. It is especially important to eliminate prepared foods containing milk purchased from the supermarket and dishes from restaurants that have sauces.**

A nine day old newborn was brought to the emergency room with severe dehydration and watery diarrhea. His temperature was 37.6 degrees and his weight was found to be significantly less than his birth weight .His mother said that the watery diarrhea started with the first feed of breast milk . The stool examination showed no pus cells and PH was 4 (normally infant PH is 5 - 5.5 ).

1- What is the most probable diagnosis for this case?

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2- What do you think the cause of lower fecal pH (5.0–5.5) in infants compared with older childrenand adolescents?

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3- What do you think the cause of lower than normal fecal PH in this case?

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4- Mention 2 other investigations to help diagnose the condition.

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5- How can you treat this case during infancy period ?

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ANSWER:

1- Congenital Lactose intolerance.

The newborn showed watery diarrhea which started after the first feed of breast milk which contains lactose .The lactose was not hydrolyzed in the small intestine due to deficiency of the intestinal lactase and went to the colon where part of it was splited by the colonic bacteria into glucose and galactose . The unsplit lactose in the colon and its fermentation products drawn water into the colon (by osmosis) leading to watery diarrhea.

The continuous watery diarrhea after each breast milk feed for nine days caused severe dehydration which caused low grade fever due to stimulation of the vasomotor centre which caused V.C. of blood vessels including skin blood vessels leading to decreased sweating and low grade fever.

Also the inability of the new born to hydrolyze lactose to obtain glucose and galactose which are sources of energy to human body lead to decreased weight of the newborn baby.

Absence of pus cells in stool together with absence of high grade fever let us think away from the diagnosis of bacterial or viral gastroenteritis.

Acidic stools more than normal for age can be explained by deficiency of lactase. The unabsorbed lactose entered the colon and was splitted into glucose and galactose. Some of the glucose and galactose were broken down by the bacteria into acids, for example, lactic acid. which turned the stool acidic.

It is a congenital type as the manifestations started with the first breast milk feed(The congenital type starts shortly after birth as human milk or lactose-containingformula is introduced).

2- Because of the physiologic overload of lactosein their diets. This in turn helped to favor growth of Lactobacillus species in the colon.

3- As already said above, the lactase deficiency caused the unabsorbed lactose to go into colon where it was splitted into glucose and galactose. Some of the glucose and galactose were broken down by the bacteria into acids, for example, lactic acid which turned the stool acidic.

4- A) Elimination diet test.

B) Hydrogen breath test

5- The infancy period means the first 2 years of baby's life.

-- In the first 4 or 6 month period where the infant is completely dependent on milk for feeding, lactose free milk formula is used.

-- At 4 or 6 month and after, on introduction of other foods ,continue with lactose free milk formula and lactose free diet is introduced. Use of partially digestedproducts (such as yogurts,cheeses, andpretreated milks **with lactase)**. Substitutes for cow milk based on rice, soy,or other proteins can be used.Also Ca and vitamin D are given.

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A 10 years old child came to the pediatric clinic with his mother and he was complaining of intestinal bloating,gases ,cramps and diarrhea following a meal containing lots of Dairy products. He said that he was complaining of chronic abdominal pain since about 5 month and his mother noticed it to occur if he increases the amount of milk he drinks more than 1 glass / meal or eats lots of ice cream. Heweighted normal for age.and eats well balanced diet. Elimination diet test and hydrogen breath test were performed and confirmed the diagnosis of lactose intolerance. Also stool examination revealed no pus cells or parasites. Immune related enteropathies were also excluded.

1- The type of lactose intolerance in this case is most probably…………………………………………….

2- Treatment of this case include……………………………………………………………………………………………

3-Which of the following dairy products you recommend which most likely cause no further problems in the future? Explain why?

a- Skim milk

b- Yogurt

c-Condensed milk

d- Ice cream

**ANSWER**:

1- Primary lactose intolerance due to the following:

--As the condition did not start after the first milk feed ,this excludes the congenital type , so the case is either 1ry or 2ry type

-- 1ry type is the most common type

-- Normal stool exclude acute bacterial infection or parasitic infestations.

-- Immune enteropathies were excluded.

. Heweighted normal for age.and eats well balanced diet excludes malnutrition

So most probably it is the primary type

2- Treatment of the case:

-- reduction of dairy products in diet to the level it cause no symptoms.

--Small amounts of lactose spaced throughout the day and consumed with otherfoods may be tolerated with no symptoms.

**-** Use of partially digesteddairy products (such as yogurts,cheeses, andpretreated milks **with lactase).** Aged cheeses may also be better tolerated.

--Substitutes for cow milk based on rice, soy,or other proteins are readily available and are generally freeof lactose.

--Oral lactase-replacement capsules ( Use of these capsules or predigested milk or dairyproducts with lactase will often permithim to be able to take some or allmilk products freely.

3- b- yogurt, because the bacteria in the yogurt contain lactase that partially digest the lactoseinto glucose and galactose before consumption.In addition,yogurt’s semisolid state slows gastric emptying resulting in fewer symptoms of lactose intolerance.