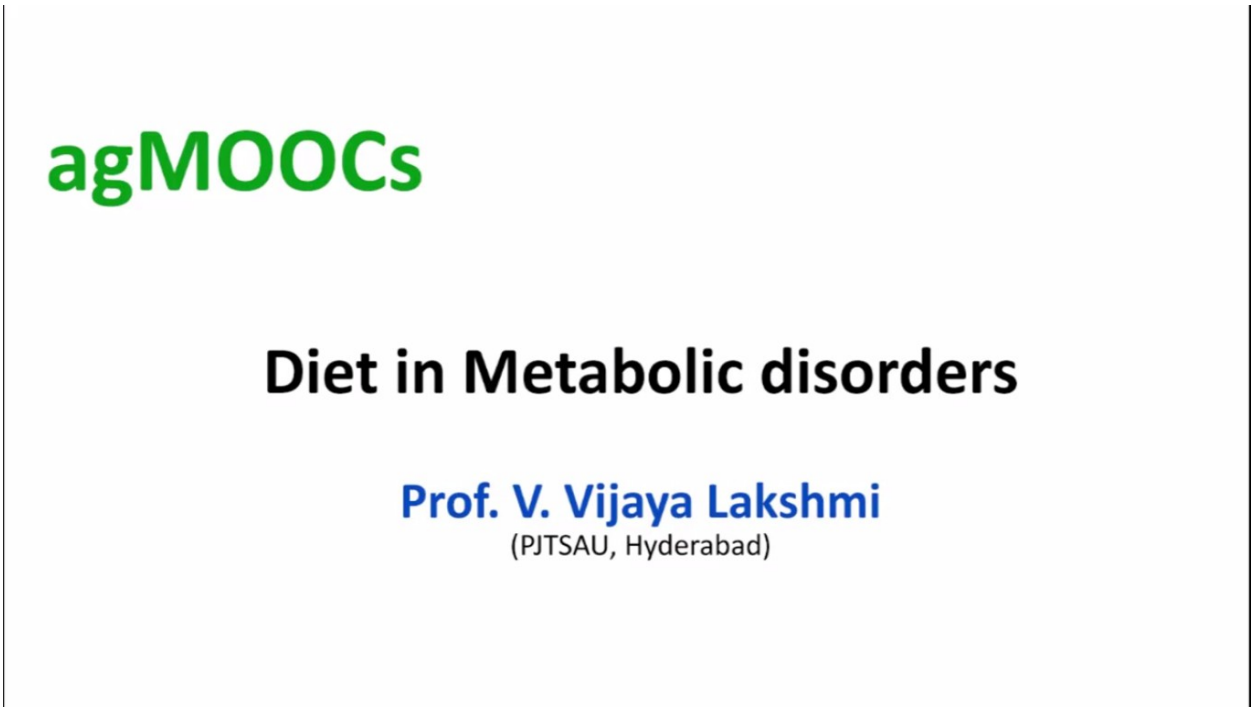

Diet in metabolic disorders

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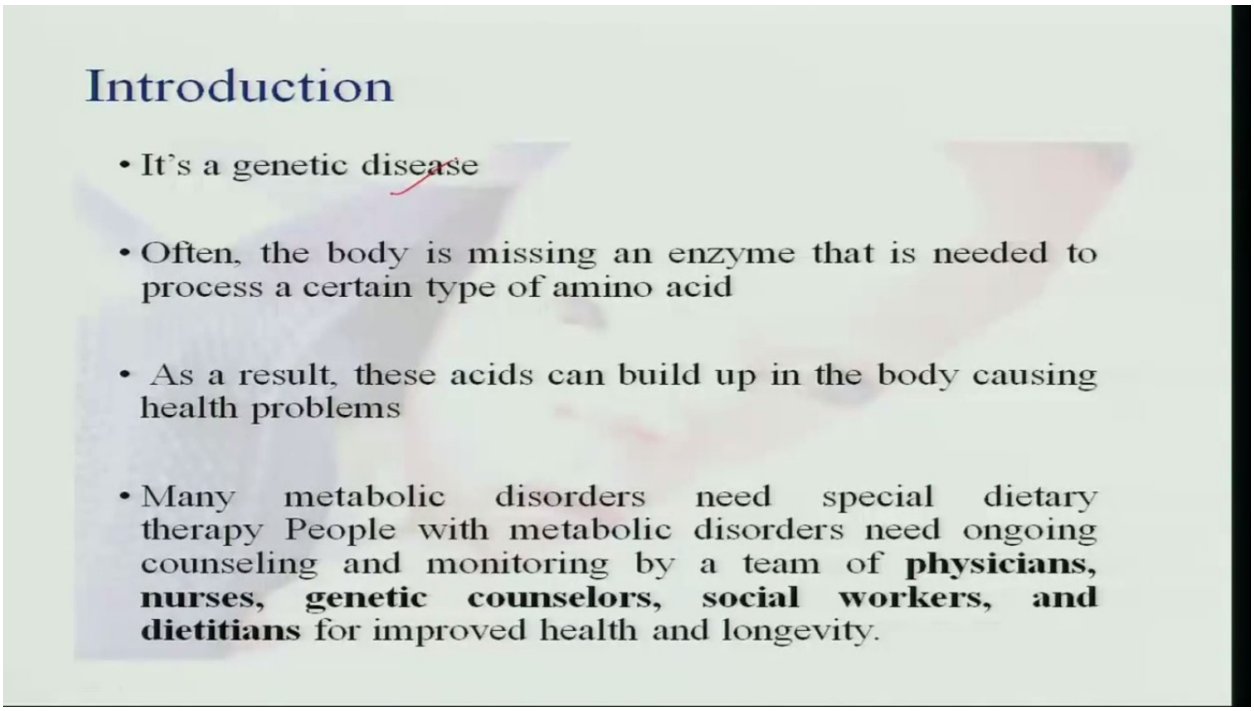
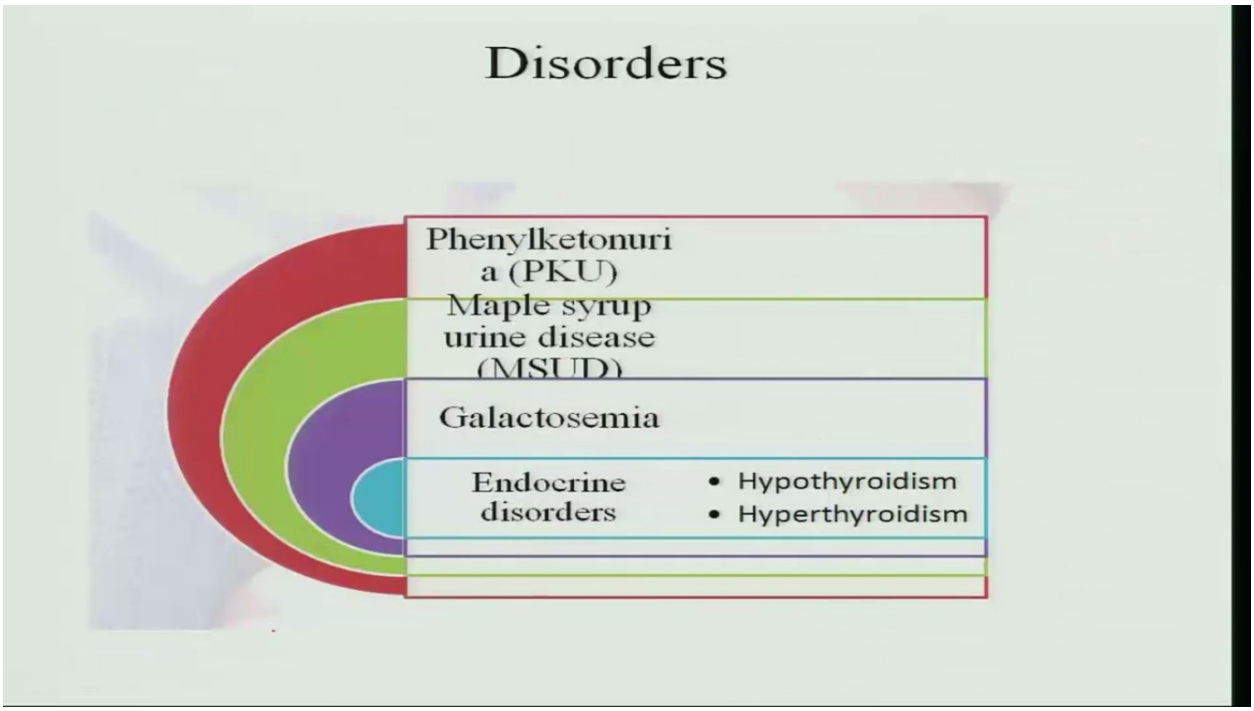
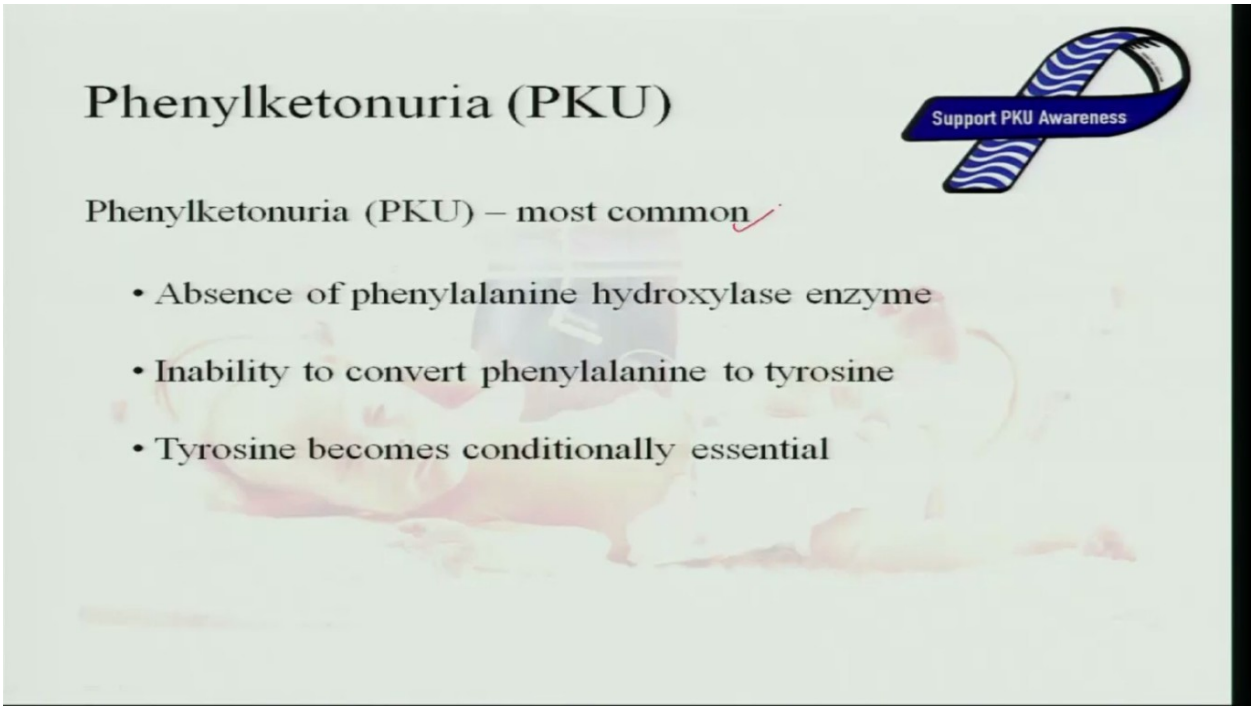
Today we will be seeing the diet in metabolic disorders. There are some disorders when the metabolism is not proper for certain amino acids or certain parts of the monosaccharides which causes a lot of problem in an infant. So these have to be recognized and suppose the particular substance or nutrient is avoided the child is safe.



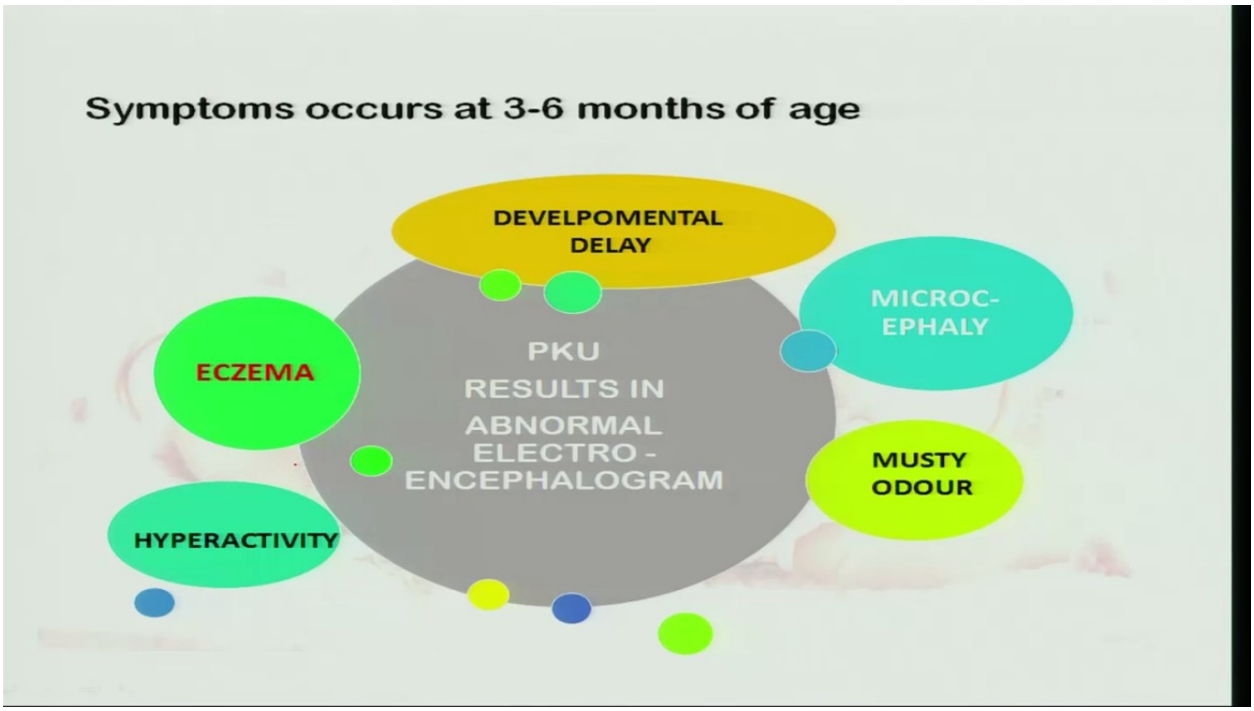
So this inborn error of metabolism it is a genetic disease. So often the body is missing an enzyme to digest a particular nutrient or a part of the nutrient. So it can be amino acid or it can be monosaccharides. So as a result the acids can build up in the body causing health problem. So there are many metabolic disorders which require special dietary treatment. So people with metabolic disorders need they have to be monitored by counseling and they have to be with a team of physicians, nurses, genetic counselors, social workers, and dieticians so that they can improve their health and also manage their health for longevity in life.



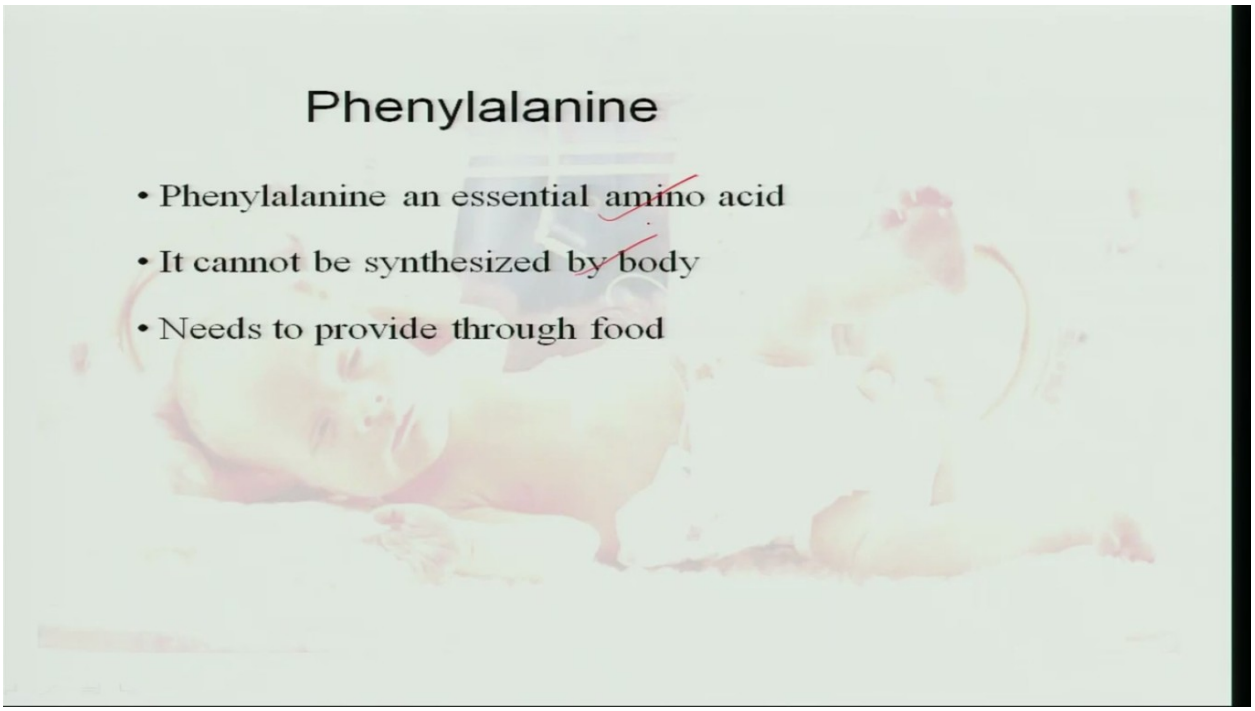
Now the disorders of the metabolism are one is phenylketonuria which is generally seen on the labels as PKU. Maple syrup urine disease that is MSUD. Galactosemia and there are endocrine disorders especially hypothyroidism and hyperthyroidism. We have been dealing with the hypothyroidism and hyperthyroidism in our earlier classes also.



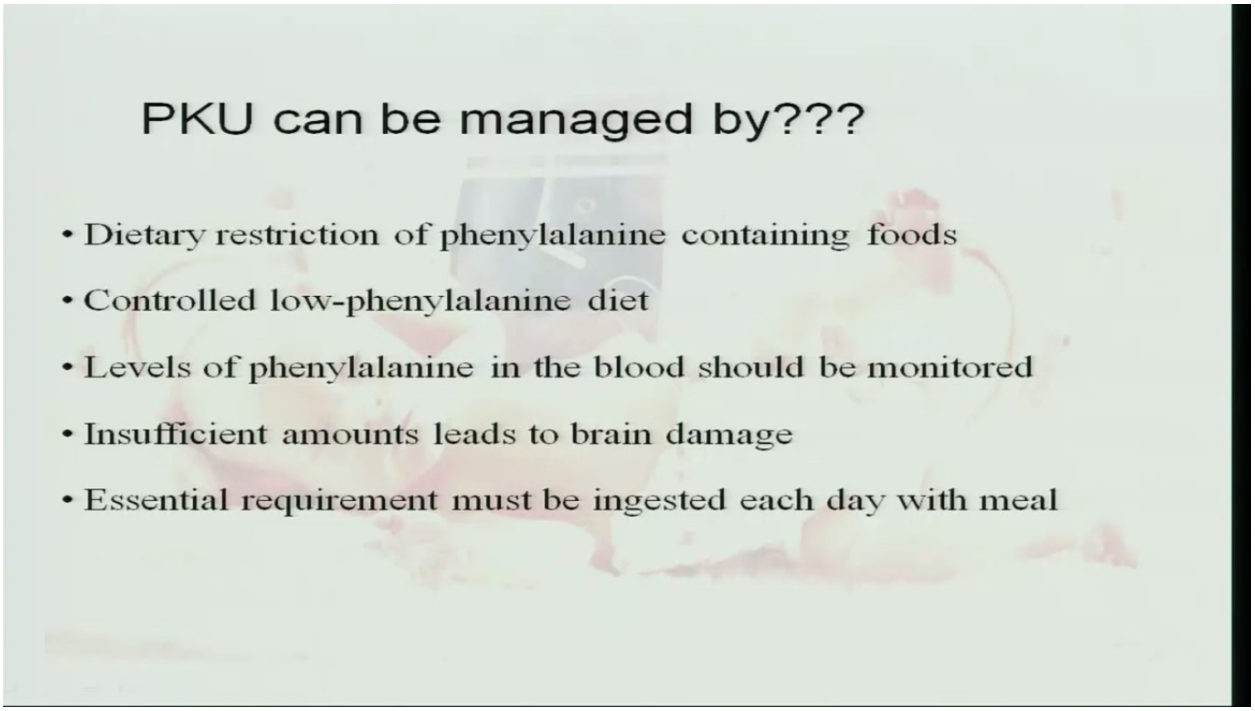
So let us see what is phenylketonuria. Phenylketonuria is the most common metabolic disorder and the absence of the phenylalanine hydroxylase enzyme in an infant causes the phenylketonuria. So it is the – this enzyme is absent and it does not convert phenylalanine to tyrosine. So the tyrosine becomes a conditionally essential amino acid for our body. So this is not a well for the body if phenylketonuria is there.



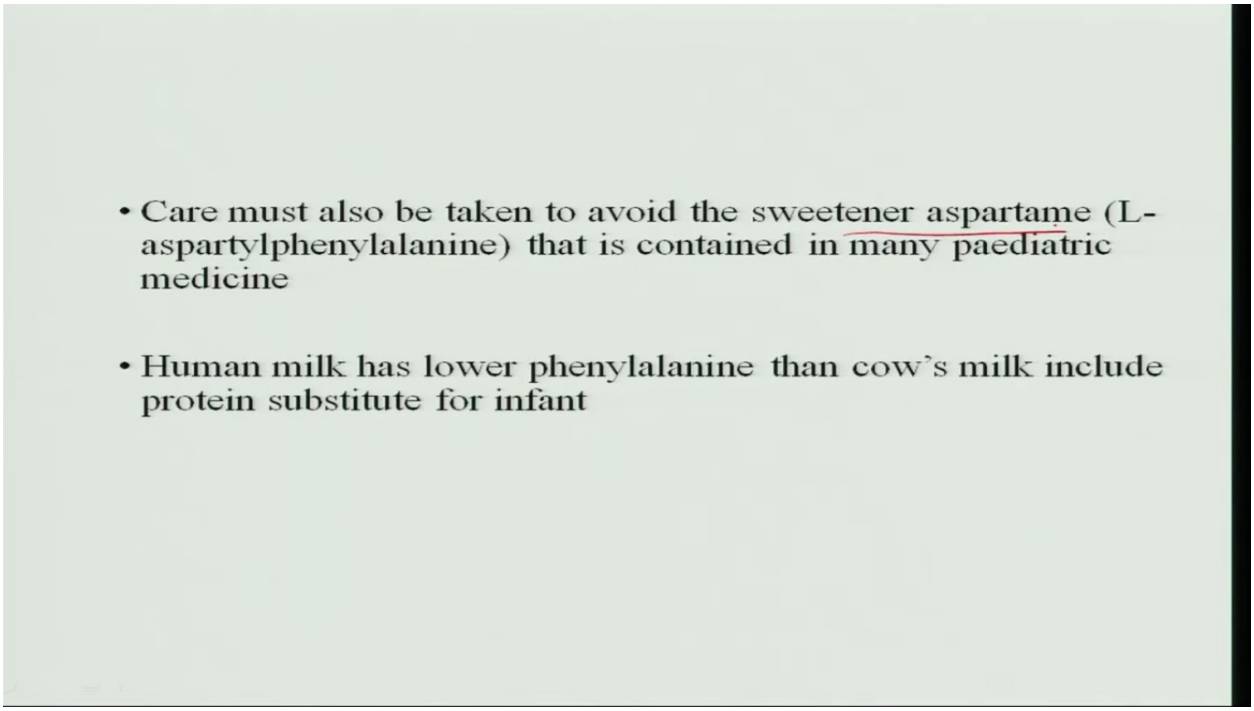
So symptoms that occur in the 3 to 6 months of age. Generally these are seen when the baby is born itself. So the child is hyperactive then it has eczema and there are developmental delays there are so many milestones a child undergoes at every stage of life. In the first year of life it learns so many activities very fast. But these are delayed and they have microcephaly that means the size of the head is very small and they have the child has a musty odour. You can find musty odor from the child. So this results in abnormal electro encephalogram that means when you test the brain activity there is an abnormal encephalogram found in the baby.



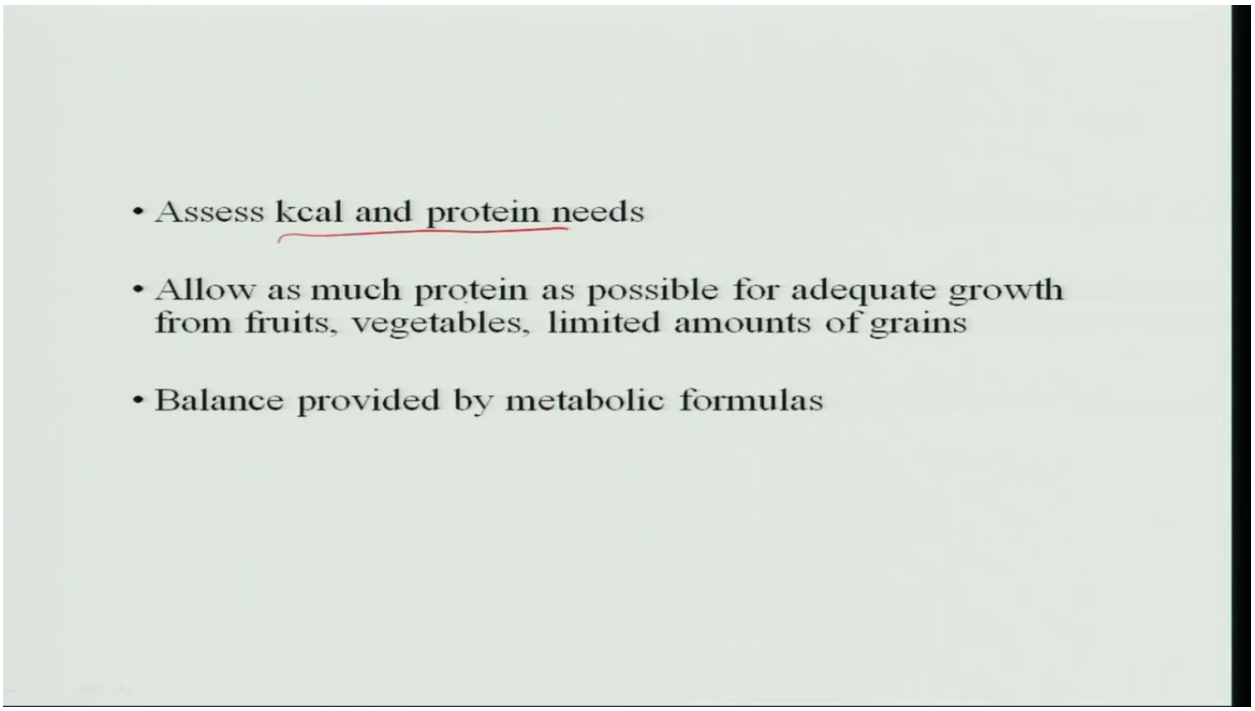
Now phenylalanine it is we have seen that it is one of the eight essential amino acids and this cannot be synthesized by the body so we have to provide the phenylalanine through food.



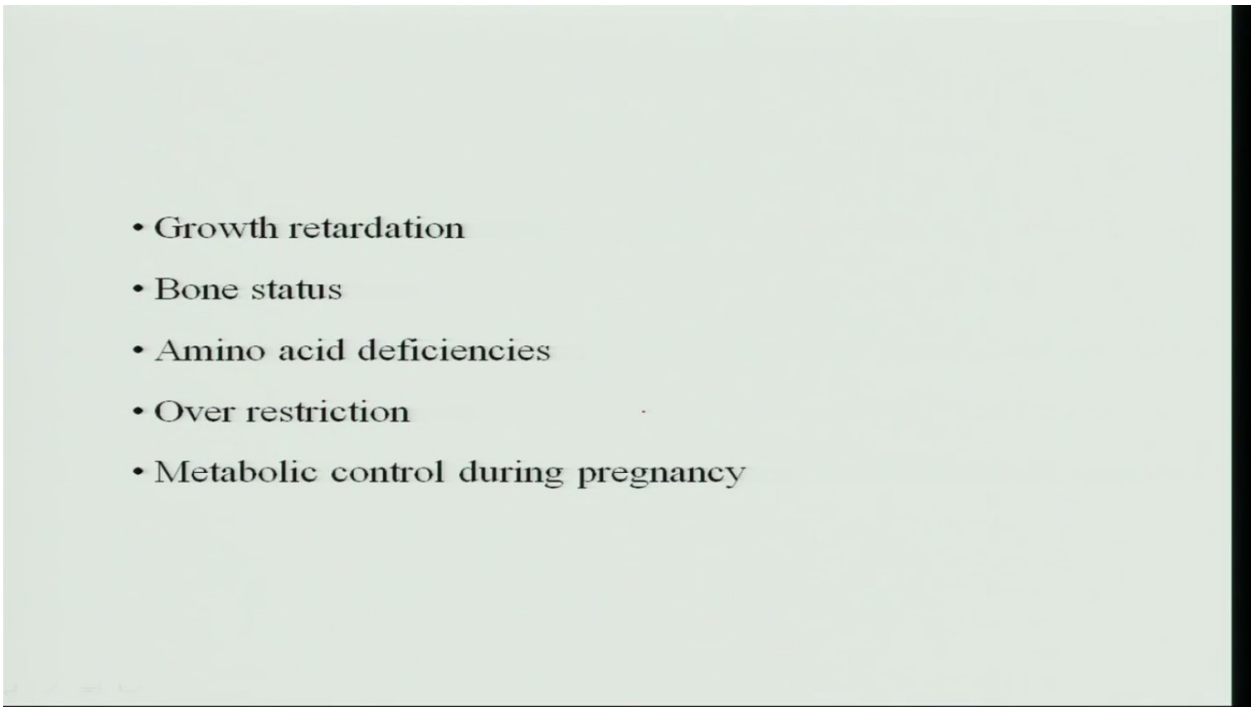
So how to manage this phenylketonuria? There should be dietary restriction of the phenylalanine containing food. Anyhow this is not synthesized in the body so it should not be given from the food. Then it can be controlled by low phenylalanine diet and the levels of phenylalanine in the blood should be monitored and insufficient amounts in the blood leads to brain damage. Now essential requirement must be ingested every day because it is an essential amino acid unless phenylalanine is there growth will not occur properly. So limited amount whatever is required by the body should be supplied to the body.



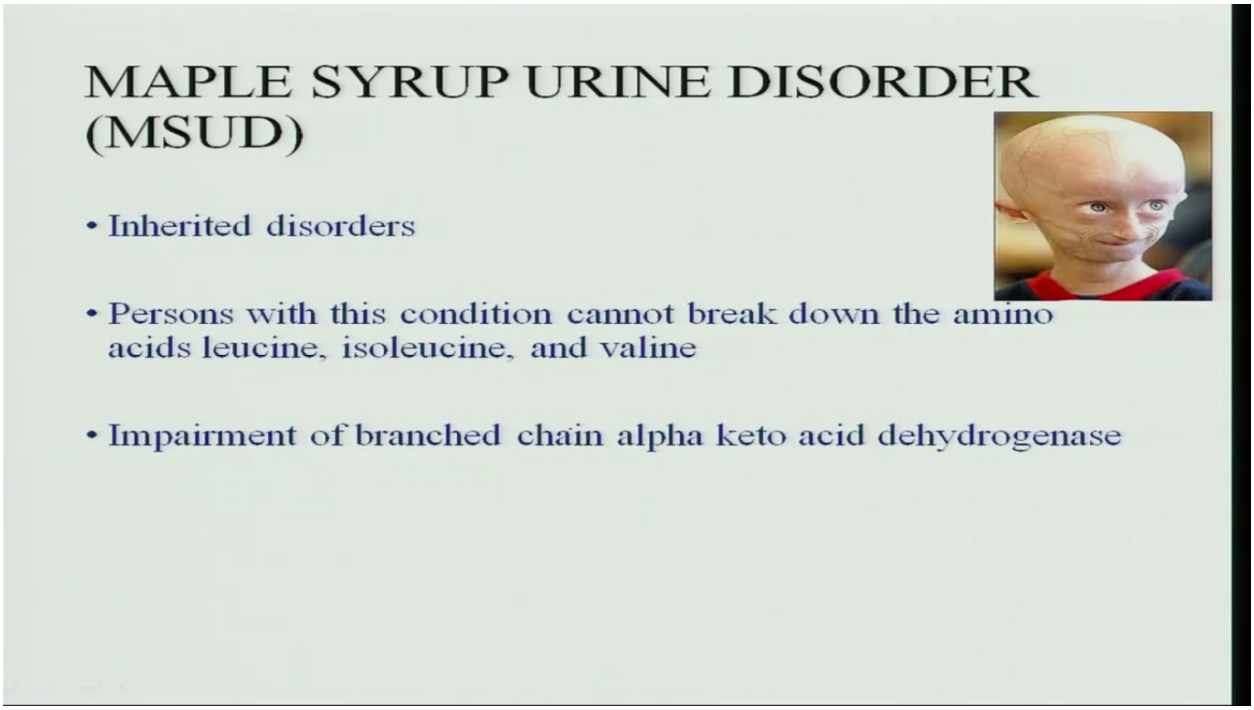
And care must also be taken to avoid the sweetener aspartame. Aspartame is a substance which acts as a sweetener and this contains phenylalanine unit. So this aspartame is added to many of the pediatric medicines/ so whenever pediatric medicine is given the label should be seen and see if it contains aspartame then it should be avoided. Then human milk has low phenylalanine compared to cow's milk therefore it should be given as a protein substitute for infant.



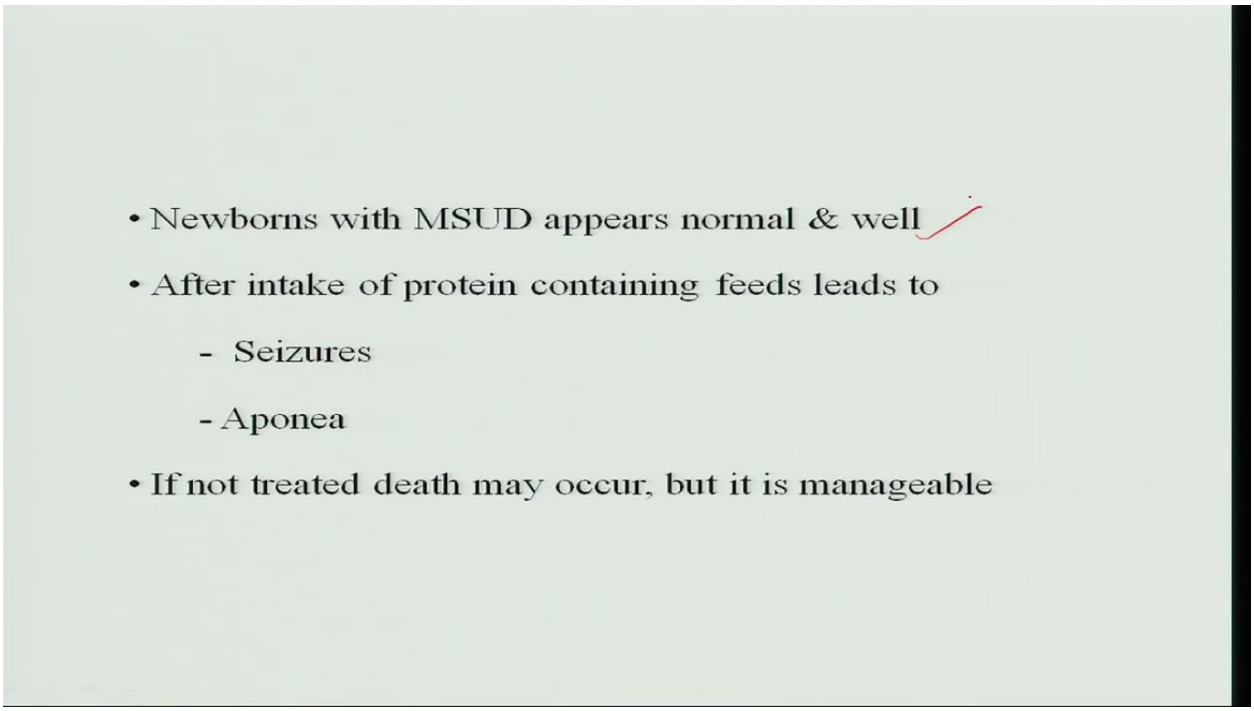
Then you assess the kilo calorie and protein intake or needs of the baby and allow much protein as possible for adequate growth. Unless protein is there there is no growth pace and infancy is the period where the highest growth in the life occurs. So you can give them from protein vegetables and limited amount of grains. So balance should be provided by the metabolic formulas. These children get very different formulas for feeding them.



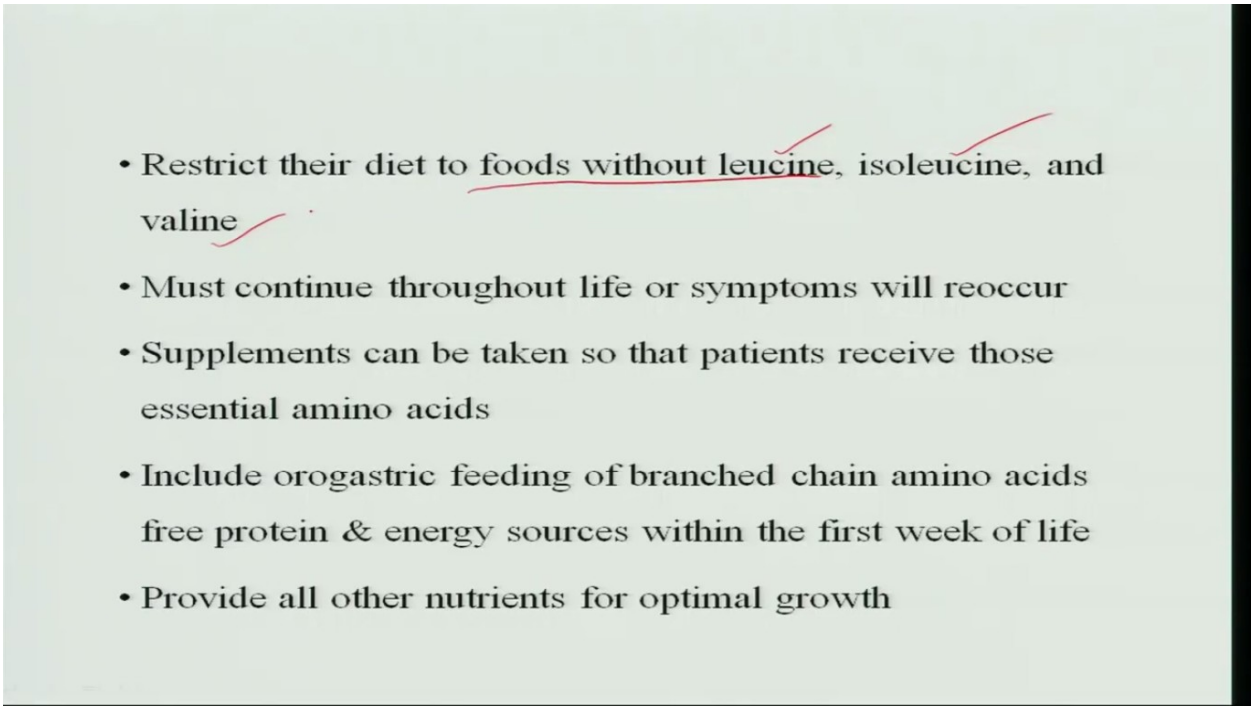
And there is growth retardation. So you have to initiate the growth in the baby. The bone status also is affected. Then the child can find many amino acid deficiency. So if you over restrict the protein also then the growth is hampered and growth does not happen normally and so metabolic controls should start during pregnancy.



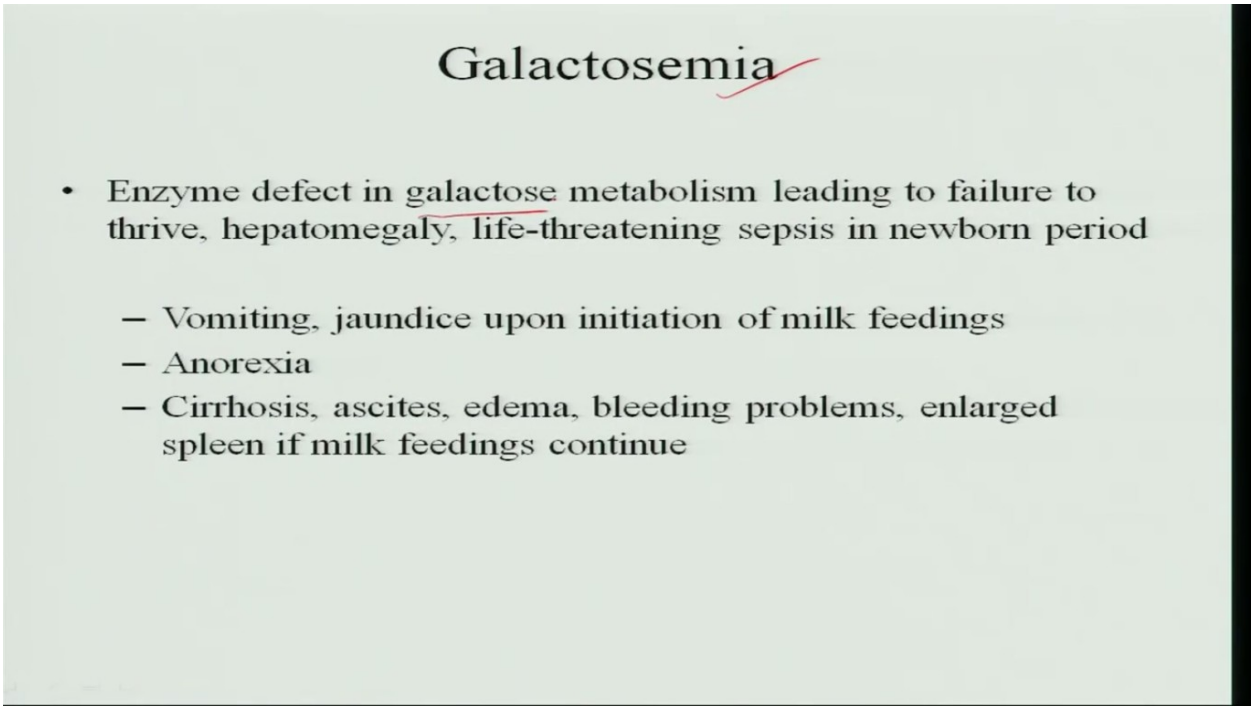
Now Maple Syrup Urine Disorder is another metabolic disorder which is again an inherited disorder. You can see the child who is suffering from the Maple Syrup Urine Disorder. So person with this condition cannot break down the amino acid, leucine, isoleucine and valine and these are very well presented in the corn and zuar. So impairment of branched-chain alpha-keto acid dehydrogenase this enzyme is not present in the child.



Newborns with the MSUS appear normal and well you cannot find that they have a deficiency but after intake of protein containing the feeds with leucine, isoleucine and valine you can find seizures in them and apnea. Apnea is they cannot breathe. So cessation of breathing or stopping of breathing occurs. So if you immediately don't see the condition and stop feeding that formula death may occur.



So restrict their diet to foods without these amino acids that is isoleucine, leucine and valine and this must continue throughout life till the symptoms appear or otherwise if you whenever you give these amino acids the symptoms will reoccur. So lifelong there is a treatment where you can – you have to avoid leucine, isoleucine, and valine and supplements can be taken so that patients receive these as essential amino acids. Now include orogastric feeding. It can be directly through the oral or directly into the gastric feeding of branched chain amino acids. Then you can give them free protein and energy sources within the first week of life. So immediately after the baby is born you can recognize these symptoms and give it a branched chain amino acids into the diet. Then provide other nutrients that are optimal for growth. Only the leucine, isoleucine, and valine are restricted but other nutrients can be given as normal.



Now Galactosemia is another metabolic disorder. Here the galactose metabolism is affected and galactose is present in milk protein. The milk sugar contains galactose. Milk contains lactose which on hydrolysis gives you galactose. And this galactose metabolism is affected and leading to failure to thrive. It leads to hepatomegaly the liver gets enlarged and there may be life-threatening sepsis that is infected which is called as sepsis and the child has vomiting, jaundice upon initiation of milk feedings, the moment you feed milk to the baby there is jaundice, anorexia. Then the child has cirrhosis, ascites, edema, bleeding problem, and enlarged spleen if the milk feedings continue. So there is a damage to all the organs in the body if the feeding is continued for a child suffering from galactosemia. They lack an enzyme called Galactus which cannot digest the galactose in the milk.

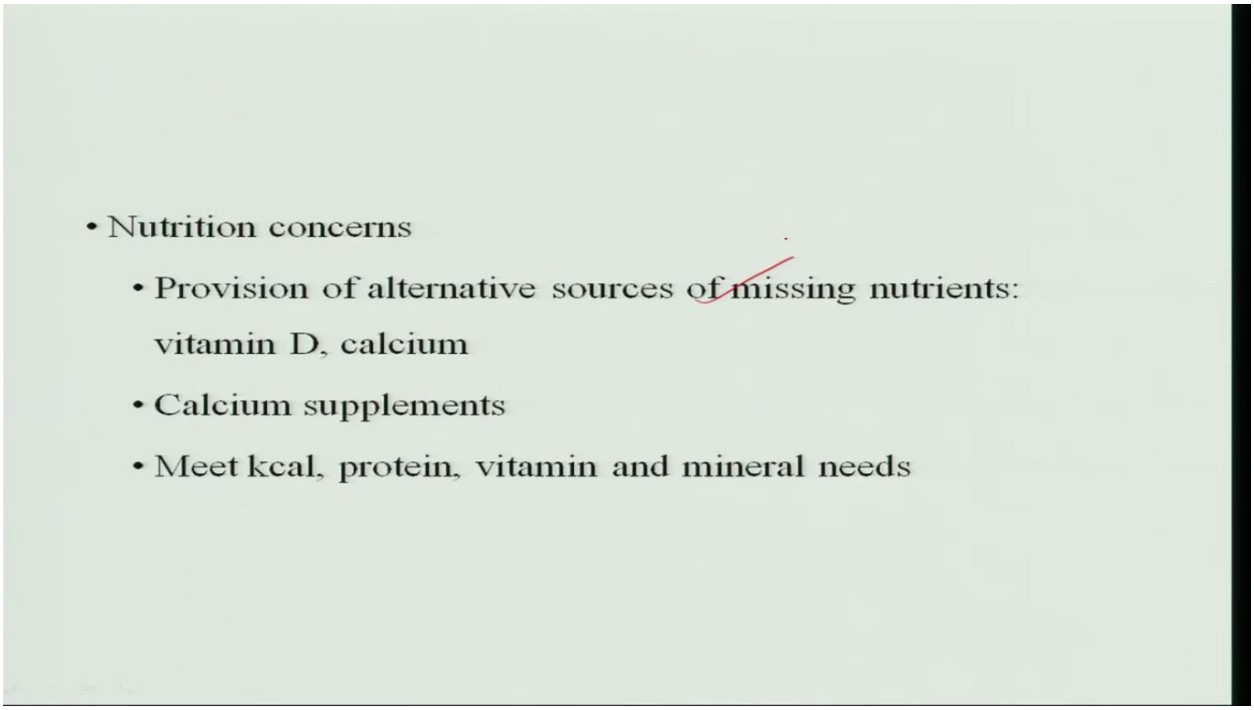
- Patients with galactosaemia are unable to metabolise galactose, most frequently due to a deficiency of the enzyme galactose-1-phosphate uridyl transferase

And patients with galactosemia are unable to metabolize galactose and they frequently fall sick and the brain growth also occurs because of galactose. There is less brain growth in such babies.

Nutrition Interventions

- Exclusion of galactose/ lactose from diet
- Immediate reversal of symptoms results
- Exclusion of human milk, cow's milk …
- Substitution of casein hydrolysate-containing formula
- Infant soy formulas

Now nutrition intervention is exclusion of galactose or lactose from the diet. The child should not be fed with milk. It can be either human milk, cows milk, or buffalo milk any type of milk contains lactose. So the milk should be avoided completely in the diet of the infant but we know that the milk is the main food for an infant. So if you feed milk there is immediate reversal of the symptoms. So exclusion of human milk cows milk can be done and substitution can be done by casein hydrolysate containing formulas because casein is the protein that is present in the milk. So we can supplement with casein and soy formulas are available. The soy milk is made into an infant soy formula and that can be fed to the baby.



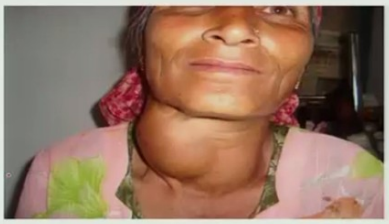
And provision of alternative sources of missing nutrients. So they have to be supplemented like vitamin D and calcium or you can find sources where vitamin D and calcium are fortified into the formula. Then calcium supplements have to be given. Then calories have to be met. Protein have to be met. Vitamins and mineral needs have to be met to the child.

Hyperthyroidism

- Is a disturbance in which there is an excessive secretion of the thyroid gland with a consequent increase in the metabolic rate

Symptoms

- Weight loss
- Excessive nervousness
- Prominence of the eyes
- Enlarged thyroid gland
- Appetite is often increased
- Weakness



Now hypothyroidism is a hormonal disorder where again the metabolism is affected. Thyroid hormone is such that it affects the energy metabolism in the body. So there is excessive secretion of thyroid gland in hyperthyroidism therefore it increases the metabolic rate almost by 100% so if the metabolic rate increases by 100% there is increased energy expenditure leading to weight loss, excessive nervousness, prominence of the eyes. Then enlarged thyroid gland you can see the thyroid gland. Then appetite is increased because the person feels very hungry as the energy expenditure is very high. Then weakness.

Modification of the diet

- High calorie (4000-5000)
- High protein diet (100-125g)
- Include snacks between meals
- Multivitamin mineral supplements are often given
- Avoid Caffeine containing foods

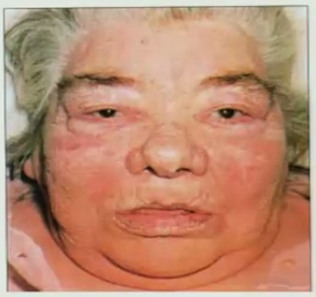
Now modification of diet can be the person has to be given since the metabolism increases by 100% you have to give the double the quantity of energy to the patient. So 4,000 to 5,000 kilocalories per day is the requirement for a hypothyroid patient. Then protein diet also has to be given more because there is lot of muscle wastage the energy is being utilized so protein reserves also are utilized for energy. Now you give them 100 to 125 grams of protein and include snacks between meals because this high amount of calorie and protein cannot be consumed in three large meals. You have to include more number of meals. So you can give in the form of snacks and fluids in between meals. Then multivitamin mineral supplements are often given. And caffeine containing foods can be avoided because they hinder the absorption of certain nutrients.

Hypothyroidism

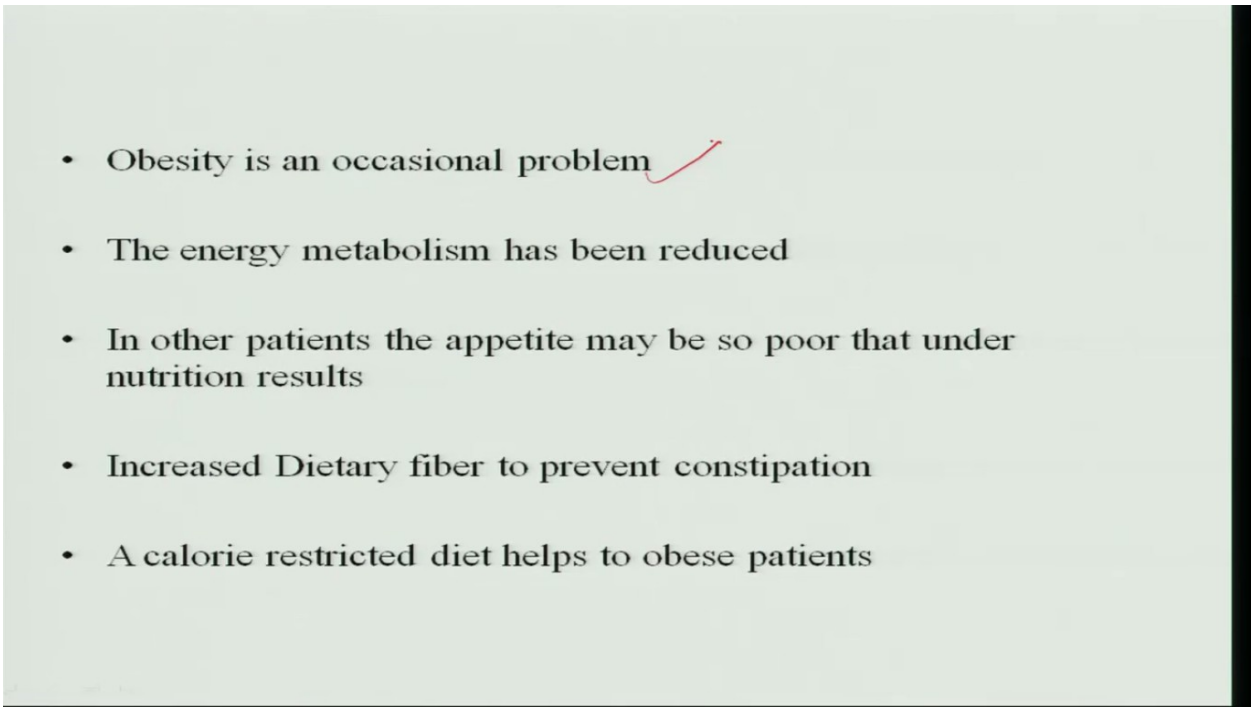
- Decreased production of the thyroid hormone is known as myxedema
- Myxedema is characterized by a lowered rate of energy metabolism 30-40% below normal

Symptoms

- Muscular flabbiness
- Puffy face, eyelids & hands
- Sensitivity to cold
- Personality change to apathy & dullness



Now hypothyroidism is where the production of thyroid hormone is decreased. So this condition is called as myxedema and when the metabolism is decreased it is almost decreased by 50% of the normal metabolism. So what happens is the whatever energy intake is there by the individual it becomes excess. Therefore the energy requirement becomes very less for an individual but at the same time we cannot give low energy foods because the individual will become weak. So we have to see the symptoms like muscle flabbiness occurs, puffy face, and eyelids and hands become puffy and sensitivity to cold they cannot all hot or cold climates, personality changes to apathy and dullness because the personality changes like this they lose interest in their surroundings and they become very dull.



Obesity becomes a problem since the energy metabolism has reduced. So some patients lose their appetite also. The appetite becomes very poor and so as a result under nutrition results. And increased dietary fiber has to be given to such patients because constipation becomes a problem as there is sluggish peristaltic moment also in the gastrointestinal system. Now a calorie restricted diet helps the obese patients to come back to their normal body weight.

Therefore these metabolic disorders have to be recognized early and the patient has to be given accordingly what type of diet he has to give and treat the patient. Otherwise, all these disorders may become fatal.

Thank you.