Diabetic with Lipodystrophy

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Abstract

Background: 18 yr-old Canadian female soccer player presented to pre-participation examination with diagnosed Type 2 Diabetes secondary to Congenital Generalized Lipodystrophy. Athlete stated she was diagnosed with Diabetes in July and Lipodystrophy at 6 months of age. Her medical records indicated at her 4-year old appointment, her insulin levels at the time were elevated at 116; and presented with acanthosis nigricans in her neck and axillac. At her 5-year old check-up, she was diagnosed with hypertrophic cardiomyopathy with trace tricuspid regurg and pulmonary insufficiency. The following year, her triglyceride levels were elevated at 2.58 and her cholesterol/HDL ratio was elevated at 4.2. At age 7, she reported with polydipsia and polyuria. Her insulin levels increased to 203 and her triglycerides levels remained elevated. She also was diagnosed with normocytic anemia with decreased levels of hematocrit; presentation of acromegaloid on the head and neck, muscular hypertrophy and phlebomegaly. At the age of 8, she showed mutations in her genetic testing of AGPAT2 gene associated with Type 1 lipodystrophy. Blood work showed increased insulin to 264. At the age of 9, she had surgery for an umbilical hernia repair. At 10 years old, she presented with venous prominence. Her bone age reports 12 years old at the age of 10. Her blood work showed her glucose levels to be 5.2 and her triglycerides level at 1.58 Athlete was discharged from the clinic at age 11. Her recent blood work for glucose screening revealed her hemoglobin levels low at 116 leading to her hematocrit levels to be low at 0.35. Her glucose was elevated at 28 and her CK was elevated 543. Her triglyceride level was high at 3.88 and her cholesterol was 5.06. Her doctor prescribed her metformin at 500mg and lantus at 22 units and Humalog at 10-12 units. **Differential Diagnosis:** Congenital Generalized Lipodystrophy with Type 2 diabetes, Centrifugal lipodystrophy, prediabetic **Treatment:** She consulted with team nutritionist and developed a diabetic diet for athletes. She was given a meal plan consisting of 6 meals a day with 100-120 g of protein and 85g of carbohydrates a day. Her glucose level should be 8-10 after meals. She is to log her food every week and report back to the nutritionist for the first month to make sure she is on track. Once a pattern is set with her eating habits, she will only log her food on game days. After a month of just game day logs, if she feels comfortable with her nutrition habits, she does not need to log her food. **Uniqueness**: Congenital Generalized Lipodystrophy is an inherited autosomal recessive disorder. This disorder is characterized with lack of fatty tissue leading to fat storage in other parts of the body. Abnormal fat storage can lead to variety of medical concerns including high levels of fats circulating in the body and insulin resistance. Athlete's doctors report she has been doing much better with management and progression than most individuals with lipodystrophy. **Conclusion**: Athletes with diabetes have to keep in mind the importance of glucose levels due to the consequences of hyperglycemic episodes and other factors leading to diabetic coma if levels are not within normal limits. Also, learning about lipodystrophy is important due to one of the correlating factors being hypertrophic cardiomyopathy, the most leading cause of cardiac death in youngathletes.

Introduction

Congenital generalized lipodystrophy (also called Berardinelli-Seip congenital lipodystrophy) is a rare case identified by a relatively complete absence of fatty tissue in the body and a very muscular body figure. Fatty tissue, also known as adipose tissue is located in various regions of the body, including underneath the skin and covering the internal organs. Adipose tissue is responsible for storing fat for the use of energy and also contributes cushioning for internal structures. A lack of adipose tissue provokes the storage of fat to move into the liver and muscles, which can lead to serious health issues. Mutations in the *AGPAT2*, *BSCL2*, *CAV1*, and *PTRF* genes produce congenital generalized lipodystrophy. The proteins created from these genes are significant in the production and activity of adipocytes. Adipocytes act as the fat-storing cells in adipose tissue. Mutations in any of these genes decrease or remove the activity of their proteins. Lack of these proteins causes damage the production, structure, or activity of adipocytes and causes the body to unsuccessfully store and use fats properly. These abnormalities of adipose tissue interrupt hormones and alter many of the body's organs, leading to the various signs and symptoms of congenital generalized lipodystrophy.

The signs and symptoms of congenital generalized lipodystrophy are generally noticeable at birth or early childhood. Due to the lack of adipose tissue and exceeded muscular tissue, individuals appear very muscular. Veins present prominent due to the lack of adipose tissue underneath the skin. Those who are affected by lipodystrophy have a large chin, prominent orbital ridges, large hands and feet, and a prominent umbilicus. One of the most common factors is insulin resistance. Insulin resistance occurs when the body's tissues are incapable of recognizing insulin. Insulin is a hormone that is needed to help regulate blood sugar levels. Insulin resistance can lead to a more serious disease called diabetes mellitus if not treated properly.

Case Report Background

Subject: 18 year-old College female soccer player with diabetes secondary to lipodystrophy participated in a single case study design

Physical Findings

Athlete stated she was diagnosed with Diabetes during PPE and Lipodystrophy at 6 months of age. Her medical records indicated at her 4-year old appointment, her insulin levels at the time were elevated at 116; and presented with acanthosis nigricans in her neck and axillac. At her 5-year old check-up, she was diagnosed with hypertrophic cardiomyopathy with trace tricuspid regurg and pulmonary insufficiency. The following year, her triglyceride levels were elevated at 2.58 and her cholesterol/HDL ratio was elevated at 4.2. At age 7, she reported with polydipsia and polyuria. Her insulin levels increased to 203 and her triglycerides levels remained elevated. She also was diagnosed with normocytic anemia with decreased levels of hematocrit; presentation of acromegaloid on the head and neck, muscular hypertrophy and phlebomegaly. At the age of 8, she showed mutations in her genetic testing of AGPAT2 gene associated with Type 1 lipodystrophy. Blood work showed increased insulin to 264. At the age of 9, she had surgery for an umbilical hernia repair. At 10 years old, she presented with venous prominence. Her bone age reports 12 years old at the age of 10. Her blood work showed her glucose levels to be 5.2 and her triglycerides level at 1.58 Athlete was discharged from the clinic at age 11. Her recent blood work for glucose screening revealed her hemoglobin levels low at 116 leading to her hematocrit levels to be low at 0.35. Her glucose was elevated at 28 and her CK was elevated 543. Her triglyceride level was high at 3.88 and her cholesterol was 5.06. Her doctor prescribed her metformin at 500mg and lantus at 22 units and Humalog at 10-12 units.

Differential Diagnosis

- Congenital Generalized Lipodystrophy with Type 2 diabetes
- Centrifugal lipodystrophy
- Prediabetic

Diagnostic Testing

- Complete Blood Count
- Physical Appearance
- Serum Concentrations of electrolytes, insulin, creatinine, triglycerides and cholesterol
- Echocardiogram,
- Skeletal survey,
- Renal & liver ultrasound.

Nutrient	*Consumption*	*Target*
Carbohydrates	343 g	340 g
Protein	266 g	400-420 g
Potassium	1838 mg	4700 mg
Calcium	563 mg	1000 mg
Vitamin C	53 mg	75 mg
Vitamin D	6 ug	15 ug
Vitamin K	72 ug	90 ug
Vitamin E	5 mg AT	15 mg AT

Results

Congenital lipodystrophy has no effect on her soccer training or physical activity. She meets with the nutritionist on site to set up a meal plan to manage her diabetes. She was advised to add more protein into her diet to slow down her carbohydrate metabolism. She eats 85 grams of carbohydrates and 100-120 grams of protein everyday. Her blood glucose levels should be at 7mg in the morning and 8-10mg after each meal. [8] Her food preferences for healthy grains are whole grain bread, whole grain cereal, and fig bars. She enjoys getting her fruits in with fresh and dried products. She also enjoys yogurt and milk. Due to her increase in protein intake, she is starting to use protein supplements to help achieve her required consumption. Once she is familiar with this amount of intake, we will start eliminating supplementation and add in high quality complete whole food proteins. Student athlete states that she is lactose intolerant so she was recommended to drink lactose free milk with her cereal instead of almond milk. She is also slightly over the carbohydrate percentage recommended by her nutritionist. She discussed with her nutritionist eating less cereal and bagels almost everyday and increasing her protein intake by eating more chicken and other meats. She is low in Vitamin C, D, E, and K. She was advised to add more fruits and vegetables.

Discussion and Summary

Lipodystrophy is a very rare congenital condition and has a lot of serious correlating factors. Diabetes is one of the most common factors of lipodystrophy. Diabetes is a abnormal rise in blood glucose levels that is also called hyperglycemia. Type 2 diabetics do not use insulin properly leading to insulin resistance. The worst of the factors is hypertrophic cardiomyopathy, which the myocardium of the heart is abnormally thick. Hypertrophic cardiomyopathy is the number 1 leading cause of sudden death of young athletes. As you can see with the case study, lipodystrophy is not a ineligible condition in sports but the relating factors can lead to serious conditions leading to ineligibly

Other possible treatments for lipodystrophy are leptin treatment has been proven profitable in managing hypertriglyceridemia and diabetes mellitus. In lipodystrophy, the leptin levels and cellular leptin sensitivity is reverse. In lipodystrophy, leptin levels are low and cells are susceptible to it; therefore, leptin replacement therapy is used as the primary treatment of choice. Currently, the 4-month leptin-replacement therapy consisting of twice-daily injection protocol was stated to enhance glucose levels and lipid metabolism in nine female patients with lipodystrophy in the United States. Considering the results of the leptin-replacement therapy, it is very certain that leptin deficiency is the primary effect of the metabolic abnormalities correlated with lipodystrophy. Under harsh discipline of changes in lifestyle and a massive high compliance of leptin injection, we establish that the leptin-replacement therapy enhances both insulin sensitivity and insulin secretion adequately and briskly improves glucose and lipid metabolism in patients with generalized lipodystrophy. Its results are managed for up to 36 months without any adverse effects. Leptin-replacement therapy is valuable to diabetic obstacles and lipodystrophic complications. The once-daily leptin injection is beneficial to manage glucose and lipid metabolism for a long period of time. Research shows that leptin-replacement therapy is a sufficient and safe treatment for long-term improvement of glucose and lipid metabolism and obstacles in generalized lipodystrophy.

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