

Berardinelli-seip congenital lipodystrophy



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Berardinelli-Seip Congenital Lipodystrophy

Congenital Generalized Lipodystrophy, most commonly known as Berardinelli-Seip Syndrome or Berardinelli-Seip Congenital Lipodystrophy (BSCL), is an autosomal recessive disorder. It affects the development of adipocytes, causing lack of subcutaneous fat and prominent musculature (Pol et al, 2015). Upon birth or during early infancy, a person with BSCL can easily be recognizable upon observation due to abnormal facies and body habitus. BSCL is a disorder that progresses as the patient grows. Mental retardation and delayed language development may also be present (Babu, Sharma, Jayaseelan & Appachu, 2008). Its progress is manifested by high levels of triglyceride and insulin resistance. By the third decade of life, more complications such as hypertrophic, stroke, cardiomyopathy, acanthosis nigricans, and diabetes mellitus may occur. The person with this disorder may also appear older than his or her age. Of all lipodystrophy disorders, BSCL is the most severe form (Mennella, & Schub, 2018).

Other than genetics, there is no other known cause for BSCL. However, a child whose parents carry the abnormal gene is at more risk. A child whose both parents carry the abnormal gene has a 25% chance of having BSCL and 50% chance of being a carrier (Mennella, & Schub, 2018). Children born from a consanguineous marriage are also at a higher risk due to a higher chance of genetic mutation (Prasad, 2011). BSCL is rare, affecting 1 in 10 million people in the United States. In Norway, it affects 1 in 1 million people. However, it is more prevalent in countries like Lebanon, Portugal, and Oman, affecting 1 in 200, 000, 1 in 500, 000, and 1 in 25, 000 people, respectively (Dantas, 2018). According to Mennella, & Schub, it is also more common of

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sub-Saharan African, Moroccan, Algerian, or Tunisian descent, as well as Middle Eastern Arabs (2018).

According to Mennella, & Schub, there are four distinct subtypes of BSCL, distinguished by their genetic cause. Type 1 BSCL is caused by AGPAT2 mutation in which the enzyme associated with is defective, causing the inability to store fats (2018). Type 2 BSCL is a mutation in the gene that encodes a protein called " Seipin" whose function is unknown but is associated with lipids droplets, (Garg, 2004). Type 3 BSCL is involved in mutation in the CAV1 gene which is responsible for the creation of the Caveolin protein which plays a role in lipid regulation (Parton, Simons, 2007). Finally, type 4 BSCL is a mutation of the PTRF gene that is responsible for the protein called polymerase I. Due to the mutation, the production of polymerase I, which also regulates the lipid, is obstructed. All gene mutation from all types affects the development, structure, and function of adipocytes (Mennella, & Schub, 2018).

Because people with BSCL cannot properly store lipids, lipids that are not stored are stay within the bloodstream which cause hypertriglyceridemia and fat deposition in organs such as liver (Lima et al., 2018). These can cause additional complication such as pancreatitis, hypertension, hypertrophic cardiomyopathy, and cirrhosis (Mennella, & Schub, 2018).

Hypertriglyceridemia is also associated with insulin resistance, causing diabetes mellitus, which is also associated with having thickened, dark skin mostly prominent in skin folds called acanthosis nigricans (Phiske, 2014; Yuan, Al-Shali, & Hegele, 2007).

Upon observation, key physical features that are almost always prominent in people with BSCL are hollowed cheeks, acromegaloid facies, abnormal body habitus, prominent musculature, and other visible lipodystrophy affecting the trunk and limbs. Acanthosis nigricans and hirsutism in girls may also be present (Dantas, 2018). Laboratory tests will indicate hyperglycemia, hypertriglyceridemia, insulin resistance, high HbA1c, and decreased serum leptin levels, to name a few. Imaging tests may also be used to monitor secondary diseases such as hypertrophic cardiomyopathy (Mennella, & Schub, 2018).

Due to multiple diseases affected by BSCL, when left untreated, new complications may occur or worsen due to secondary symptoms. By the third decade of life, hypertrophic cardiomyopathy, hepatic cirrhosis, pancreatitis, advanced bone age, diabetes mellitus, and arteriosclerosis may occur. Mental disability may occur as well as polycystic ovary syndrome in females (Mennella, & Schub, 2018). In very severe cases, unmonitored secondary diseases such as liver disease, gastrointestinal bleeding, sepsis, pneumonia, septic arthritis, diabetes, and kidney failure may cause death in later years (Lima et al., 2018).

Blood tests are performed to monitor affected systems. Metabolic panel tests are used to determine levels for hyperglycemia. Serum lipid tests are used to monitor triglycerides. Serum liver enzymes are tested to determine if the patient developed steatosis or cirrhosis, and serum lipase and amylase are tested to monitor for pancreatitis. Genetic testing is also used to identify what type of BSCL the person has. Other diagnostic tests include CT scan to look for the absence of adipose tissues, as well as to look for abnormalities in <https://assignbuster.com/berardinelli-seip-congenital-lipodystrophy/>

the livers. MRI of the bones is done to look for the absence of fat in the bone marrow, subcutaneous, and intramuscular fat. X-rays are used to scan the extremities to look for signs of elongated bones and cystic lesions.

Echocardiogram and EKG are used to monitor for hypertrophic cardiomyopathy. Finally, bone density may also be tested to assess for osteopenia or osteoporosis (Mennella, & Schub, 2018).

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