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Familial Partial Lipodystrophy: Short Communication

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Short Communication

Familial partial lipodystrophy (FPL) may be a rare genetic disease characterized by selective, progressive loss of body fat (adipose tissue) from various areas of the body. Individuals with FPL often have reduced subcutaneous fat within the arms and legs and therefore the head and trunk regions may or might not have loss of fat. Conversely, affected individuals can also have excess subcutaneous fat accumulation in other areas of the body, especially the neck, face and intra-abdominal regions. Subcutaneous fat is that the fatty or fat layer that lies directly beneath the skin. In most cases, fat loss begins during puberty. FPL are often related to a spread of metabolic abnormalities. The extent of fat loss usually determines the severity of the associated metabolic complications. These complications can include an inability to properly breakdown an easy sugar referred to as glucose (glucose intolerance), elevated levels of triglycerides (fat) within the blood (hypertriglyceridemia), and diabetes. Additional findings can occur in some cases. Six different subtypes of FPL are identified. Each subtype is caused by a mutation during a different gene. Four sorts of FPL are inherited as autosomal dominant traits; one form is inherited as an autosomal recessive trait. The mode of inheritance of FPL, Kobberling variety is unknown.

Lipodystrophy may be a general term for a gaggle of disorders that are characterized by complete (generalized) or partial loss of fat. Additionally to FPL, there are other inherited sorts of lipodystrophy. Some sorts of lipodystrophy are acquired at some point during life. The degree of severity and therefore the specific areas of the body affected can vary greatly among the lipodystrophies. Some individuals may only develop cosmetic problems; other can develop life-threatening complications. The loss of fat that characterizes these disorders is usually mentioned as lipoatrophy instead of lipodystrophy by some physicians. FPL was first described within the medical literature in 1970s independently by three groups of physicians, including Doctors Ozer, Kobberling and Dunnigan.

Treatment consists of correcting metabolic abnormalities and managing complications. Monitoring diet (reduced intake of dietary fats and carbohydrates) and maintaining daily physical activity can improve the metabolic complications of lipodystrophy. Insulin sensitizers (mainly metformin) and lipid-lowering drugs (statins, or fibrates just in case of major hypertriglyceridemia) also can be helpful. Diabetes may require other non-specific treatments, along side insulin. The orphan drug metreleptin is permitted under exceptional circumstances in Europe for the treatment of metabolic complications of partial sorts of lipodystrophies, in adults and youngsters above the age of 12 years, where standard treatments have failed. Further studies are required to research the advantages and risks of treatment. Regular cardiac monitoring is suggested. Ethinylestradiol should be avoided in women with FPLD and hypertriglyceridemia. cosmetic surgery can help some patients.

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