



MILROY'S DISEASE- A CONGENITAL LYMPH EDEMA

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Abstract

The congenital diseases in children are always a broadly defined area in modern pediatrics. Yet the prompt diagnosis of congenital diseases is still a quizzical scenario. Certain diseases are still a under research and miles to get hold. One among those is congenital lymphatic edema commonly named as Milroy's Disease. In some it is seen associated with other congenital anomalies like clubfoot. The most common presentation of Milroy Disease is unilateral lower extremity swelling due to lymph edema. The severity and long term effect is difficult to predict in this condition.

Key words: congenital anomalies, clubfoot, lymphatic edema

Introduction

Milroy's disease is a familial disease characterized by lymph edema, commonly in the legs, caused by congenital abnormalities in the lymphatic system. Disruption of the normal drainage of lymph leads to fluid accumulation and hypertrophy of soft tissues. It was named by Sir William Osler for William Milroy, a Canadian physician, who described a case in 1892, though it was first described by Rudolf Virchow in 1863. Other names includes hereditary lymph edema type I and Nonne-Milroy lymphedema.¹

Incidence and Transmission

This disease is more rated in females and an association with the gene *FLT4* has been described-*FLT4* codes for VEGFR-3, which is implicated in development of the lymphatic system.

Milroy's disease is also otherwise known as primary or hereditary lymphedema type 1A or early onset lymphedema. It is a really rare condition with only about two hundred cases reported in the medical literature. Milroy's disease is an autosomal_dominant condition caused by a mutation in the *FLT4* gene which encodes of the vascular endothelial growth factor receptor 3 (VEGFR-3) gene located on the long arm (q) on chromosome 5 (5q35.3).Milroy disease is inherited in an autosomal dominant pattern, which means one copy of the altered gene

in each cell is sufficient to cause the disorder. In several cases, an affected person inherits the mutation from one affected parent. Other cases could result from new mutations within the *FLT4* gene. These cases occur in people with no history of the disorder in their family. About 10 percent to 15 percent of people with a mutation in the *FLT4* gene do not develop the condition of Milroy disease².

Features

The most common presentation of Milroy's Disease is unilateral lower extremity swelling due to lymph edema, and may also be accompanied by hydrocele. Males and females may have up slanting toenails, deep creases in the toes, wart-like growths (papillomas), and prominent leg veins. Some individuals develop non-contagious skin infections called cellulitis. This condition can destroy the thin tubes that carry lymph fluid (lymphatic vessels). Episodes of cellulitis can cause further swelling in the lower limbs congenital familial lymph edema

Diagnosis

Milroy's disease **should be suspected** in individuals with the following findings:

- Lower-limb swelling that is usually but not always bilateral, present at birth or develops soon after
- Large-caliber veins
- Up slanting toe nails

Confirmation can be done by molecular genetics and evaluation of the limb swelling by lymphoscintigraphy. Radioactive colloid is injected into the toe web spaces and uptake in the ilioinguinal nodes is measured at intervals. Lymphoscintigraphy is to confirm if there is lack of uptake of radioactive tracer.³

Management

Management can only be conservative measures. Certain treatments for lymph edema disorders may possibly alleviate specific symptoms; no cure and it is usually congenital genetic counseling can be done. May have similar health conditions, delays, disorders, and physical traits associated with other lymphatic genetic diseases and chromosome abnormalities

Management is mostly done in conservative way and usually successful in most of the individuals. The goal of treatment is to cut down swelling and halt complications. Management of lymph edema should be guided by a lymph edema therapist. Complete decongestive therapy (CDT) could be used for manual lymph drainage combined with compression bandaging, skin care, exercise, and well-fitted compression garments. Some improvement is usually possible using these methods⁴.

To prevent cellulitis, foot infections (such as athlete's foot) should be treated without delay. These problems can be prevented to some extent, and prophylactic antibiotics may be needed in recurrent cases. The individuals who are diagnosed with Milroy's disease should try

to avoid wounds to swollen areas, long periods of immobility, medications that can cause increased leg swelling. The drugs such as calcium channel-blocking agents and non-steroidal anti-inflammatory drugs can worsen the condition. The excessive salt intake should be halted in order to prevent fluid retention and edema⁴.

The symptoms and severity of Milroy's disease can vary among individuals. The affected person (even within the same family), the effects of the condition may be difficult to predict. The nature and extent of swelling varies in degree and distribution. The swelling imposed by the lymphatic edema can be disabling and disfiguring. This changes with individual outlook toward the chronic lymph edema as well as whether complications arise

Various surgical management techniques were used initially to correct the lymphatic block and edema but most of them were having less success rate in completely resolving the Milroy's disease. Hence surgical measures are least recommended.

The most helpful preventive measure for the Milroy's disease can be the following

Agents/circumstances to avoid: Wounds to limbs; long periods of immobility with the legs in an exceedingly dependent position; and medications that can cause increased leg swelling.

Evaluation of relatives at risk: Evaluating the family genogram improves the identification of risk individuals and those who will benefit from treatment early in the disease course.

Genetic counseling for parents since it is an autosomal inherited diseases.

Complications

Complications of lymph edema may include recurrent attacks of cellulitis and/or lymphangitis, bacterial and fungal infections, deep venous thrombosis, functional impairment, cosmetic embarrassment, and amputation. Complications following surgery are common. It has also been reported that people with chronic lymphedema for many years may have a considerably higher risk to develop lymphangio sarcoma (a type of angiosarcoma). This type of tumor is terribly aggressive and has a very poor prognosis⁵.

Conclusion

Milroy disease is a congenital lymphatic edema and is an inherited disease. The evaluation of family members also can help to identify the pattern of transmission in the family. Congenital genetic counseling can be a helpful tool for the family and the individual. The severity of the condition is not predictable in the individuals. The diagnosed individual must follow the safety instructions to avoid wounds in the extremities and longer periods of immobility

References

- 1.Milroy WF (1892). "An undescribed variety of hereditary edema". New York Medical Journal. 56: 505–8. 2.James WD, Berger TG, Elston DM, Andrews GC, Odom RB (2006). Andrews' Diseases of the Skin: clinical Dermatology. Saunders Elsevier. p. 849. [ISBN 978-0-7216-2921-6](#). [OCLC 937244604](#)
- 3.Strayer DL, Rubin R (2007). Rubin's Pathology: Clinicopathologic Foundations of Medicine (5th ed.). Hagerstwon, MD: Lippincott Williams & Wilkins. ISBN 978-0-7817-9516-6
- 4.Glen W Brice, Sahar Mansour, Pia Ostergaard, Fiona Connell, Steve Jeffery, and Peter Mortimer. Milroy Disease. GeneReviews. September 25, 2014; <http://www.ncbi.nlm.nih.gov/books/NBK1239/>
- 5.Rockson SG. Causes and consequences of lymphatic disease. Ann N Y Acad Sci. 2010;1207 Suppl 1:E2-E6. doi:10.1111/j.1749-6632.2010.05804.x

