

# Clubfoot: Etiology and Treatment

## Editorial Comment

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Clubfoot occurs in one in 1000 live births and is one of the most common birth defects involving the musculoskeletal system. Approximately 80% of clubfeet occur as isolated birth defects and are considered idiopathic. While it is becoming increasingly clear that clubfoot is multifactorial in origin, genetic factors clearly play a role as suggested by the 33% concordance of identical twins and the fact that nearly 25% of all cases are familial. Identifying clubfoot at the time of birth is straightforward in most cases because of its characteristic appearance and resistance to correction with passive movement. When untreated, children with clubfoot walk on the sides of their feet instead of the soles, resulting in abnormal callus formation, arthritis, potential infections, and significant limitations in mobility and employment opportunities.

Because the etiology of clubfoot remains unclear, the first goal of this month's symposium is to present a group of papers dedicated to research involved in better understanding the cause of clubfoot at the most fundamental level. This type of research is crucial as identification of the exact etiology of clubfoot may eventually be helpful in determining both prognosis and the selection of appropriate treatment methods in an individual patient. Though the exact genetic mechanism of clubfoot has not yet been determined, most evidence points to a polygenic causation where multiple genes and environmental factors are responsible. There is ongoing research to identify those responsible genes. The PITX1 gene, which encodes

a transcription factor that is expressed almost exclusively in the hindlimb, is the first gene implicated in clubfoot. It is yet to be seen what percentage of idiopathic clubfoot cases this gene mutation may explain. Future genome-wide association studies will provide the best approach for the identification of other clubfoot susceptibility genes. Collaborative efforts are necessary with this type of research to have large enough sample sizes to be able to identify both major and minor susceptibility genes if they are present.

Despite the frequency of clubfoot, there is still controversy over optimal treatment strategies. Therefore the second goal of this symposium is to present papers focused on treatment strategies for clubfoot. In particular, what are the limits of the Ponseti method? What is the role for surgery in the modern day treatment of clubfoot? A trend over the last decade has been a movement away from extensive soft-tissue release surgery for definitive clubfoot management and toward less invasive strategies focused more on manipulation and castings. The most popular less invasive strategies include the Ponseti method and the French physiotherapy method. There are both successful long-term and short-term data published on the Ponseti method demonstrating its effectiveness in achieving and maintaining correction long-term without the need for extensive release surgery in the vast majority of cases. The Ponseti method consists of serial manipulation and casting, Achilles tendon tenotomy, and foot abduction bracing. There is also success reported with the French method which requires daily manipulations, taping, and physical therapy though a higher percentage of clubfoot patients treated in this manner appear to require subsequent posterior ankle release operations. Recent reports combine the serial casting, Achilles tendon tenotomy, and foot abduction bracing of the Ponseti method with the use of physical

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therapy (French method) to help improve and maintain ankle dorsiflexion long-term. This is an area that deserves further study.

Understanding the genetic etiology of clubfoot and investigating the efficacy of treatment strategies for clubfoot are not separate areas of research. Each complements

the other. Identifying the causes of clubfoot may be helpful for prognosis, risk of comorbidities, and response to treatment. Personalized treatment based on etiology may allow reduced brace wear if risk of relapse correlates with a certain genetic profile. There is still so much to be learned and so many questions left to ask.