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Current Concept Review

Prenatal Counseling for Congenital Clubfoot

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ABSTRACT

Congenital clubfoot is a common deformity that affects 1 in 1000 newborns and is frequently detected prenatally during routine prenatal care. A wide variety of detection methods and testing are used to identify clubfoot and other congenital anomalies in the fetus, including complete ultrasonography, amniocentesis, chorionic villus sampling, or cell-free DNA. Newer studies have associated certain genome sequences to clubfoot specifically. It is important for orthopaedic surgeons to understand the implications of the various tests to provide the appropriate prenatal counseling. Early prenatal detection of clubfoot can help parents prepare for the demands of caring for a child with clubfoot and build trust between families and clinicians, ultimately leading to better, patient-centered care for their children.

Key Concepts:

(1) Prompt recognition and treatment of congenital clubfoot is imperative to facilitate optimal treatment.
(2) Prenatal diagnosis usually consists of sonography of the plantar surface of both feet at 13–16 weeks' gestation.
(3) Additional post-natal testing may be indicated in patients with neurological impairment or syndromic features.
(4) Genetic markers such as PITX1, RBM10, HOX, and CASP (among others) have been identified as involved in clubfoot development and have implications on prenatal testing and counseling.

Introduction

Congenital clubfoot occurs with a reported incidence of 1 in 1000 live births per year [1]. Untreated clubfoot can lead to lower limb disability with concurrent pain and gait disturbances [2]. Therefore, parents and caregivers are often overwhelmed when they receive a clubfoot diagnosis at the fetal survey prenatal ultrasound, typically done around 20 weeks' gestation. Clubfoot is one of only a few orthopaedic congenital abnormalities detected months before birth. Knowledge of this condition places the pediatric orthopaedic surgeon in a unique position to provide counseling prior to the time that orthopaedic care is needed. These consultations educate families about clubfoot, allay anxiety they may have about the deformity and its consequences, and prepare them for Ponseti treatment before their baby is born.

The purpose of this article is to present a framework for the pediatric orthopaedic surgeon to follow on how to counsel and support families seeking a prenatal evaluation for their unborn child with clubfoot. Additionally, different types of prenatal testing for clubfoot, specific genetic associations, and related syndromes will be discussed to facilitate a comprehensive and informative prenatal consultation.

Prenatal diagnosis

Some risk factors for clubfoot are male sex, family history, parental tobacco use, maternal obesity, lower parity, early amniocentesis (<15 weeks), lower educational level, and diabetes [3,4]. Of note, mothers under the age of 35 years and mothers who are black are less likely to have clubfoot detected prenatally [5].

Congenital clubfoot can be detected as early as 13 weeks' gestation through transvaginal ultrasound or 16 weeks' gestation through transabdominal ultrasound [6]. Prior to week 13, plantarflexion or adduction of the foot can be a normal developmental finding [7]. International guidelines recommend identifying both legs and feet in a typical second-trimester fetal ultrasound scan (description of position and relationship is not necessary) [8]. A sonographic diagnosis of clubfoot is

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based on visualization of the plantar surface of the fetal foot in the same sagittal plane as the tibia and fibula (Fig. 1) [9].

Obstetricians recommend a follow-up ultrasound to confirm the diagnosis of clubfoot. Contrary to other anomalies that are measured and require surveillance in utero such as renal pyelectasis or choroid plexus cyst (which may resolve), clubfoot does not need to be monitored with serial ultrasounds. Ultrasound has been shown to have a high sensitivity of 81%; however, the false positive rate is approximately 14%. Furthermore, in fetuses diagnosed with isolated clubfoot prenatally, approximately 11% were later found to have other somatic abnormalities after birth, and thus subsequently recategorized into complex clubfoot (according to the obstetrical classification, discussed in the next paragraph) [10]. There is no role for additional or serial ultrasounds for monitoring of clubfoot, assuming adequate images have been obtained and the diagnosis is conclusive. However, as noted above, neither accuracy nor sensitivity are 100%, and some cases of clubfoot are only diagnosed postnatally [10].

Clubfoot can be unilateral or bilateral. The incidence of a prenatal diagnosis with unilateral clubfoot is slightly higher than being diagnosed with bilateral clubfoot (53.2% vs 47.8%) [10]. Prenatally, obstetricians classify clubfoot into *isolated* and *complex* cases. It is important to understand that the definition of *complex* is different in the prenatal context compared to the use of the term in children undergoing Ponseti treatment. Ponseti introduced the concept of "complex clubfoot" in 2006 in reference to those with "rigid equinus, severe plantar flexion of all metatarsals, a deep crease above the heel, a transverse crease in the sole of the foot, and a short and hyperextended first toe" that does not respond to the standard treatment protocol [11].

Obstetricians characterize the diagnosis as isolated when there is an unknown cause and when there are no other anatomic abnormalities. A clubfoot is characterized as complex by obstetricians in the setting of a known mutation, genetic syndrome, and/or additional anatomic abnormalities identified on ultrasound (Table 1). Isolated clubfoot is more common than complex clubfoot (69.7% vs 30.2%) [10]. It is important to note that the diagnosis of bilateral clubfoot does not increase the odds of being complex.

Obstetrical prenatal counseling for clubfoot

The first counseling provided to families of a child with clubfoot will always be provided by the obstetrics team. Though clubfoot is often isolated, there may be a need for additional prenatal or postnatal testing in some cases, such as if there are signs of neurological impairment or syndromic features. When a clubfoot is detected prenatally, obstetricians follow the following algorithm:

- Complete a more detailed sonogram. This can uncover other anomalies, which may change the diagnosis to complex clubfoot. The more detailed sonogram can also reveal intrauterine abnormalities that might contribute to fetal anomalies, such as bicornuate uterus, fibroids, synechiae, and amniotic bands.
- Regardless of age, recommend/offer genetic counseling and chromosomal evaluation (karyotype and microarray).
- A fetal echocardiogram should be performed because cardiac anomalies are most commonly associated with clubfoot.
- A third-trimester sonogram may be considered to rule out polyhydramnios, which could be an indicator of a neurologic disorder.
- Additionally, the family should be referred to a pediatric orthopaedic surgeon.

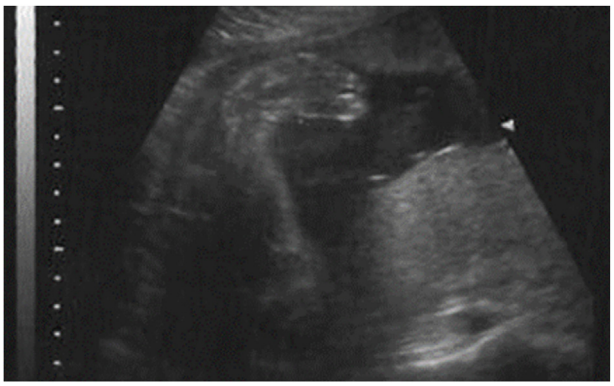

Figure 1. Two-dimensional prenatal ultrasound of clubfoot at 27 weeks.

Clubfoot can be associated prenatally with myelomeningocele, limb deficiencies, or other genetic syndromes and rare disorders such as congenital myotonic dystrophy, chromosome 22q11 deletion syndrome, and aneuploidies in which cells have additional or missing chromosomes, typically resulting from the failure of chromosomal separation during cell division (nondisjunction). Of note, Trisomy 13, 18, 21 are examples of aneuploidies with additional chromosomes. Aneuploidies can be detected in utero using both fetal genetic studies (eg, amniocentesis) and cell-free DNA screening. Mutations (duplications and deletions) of chromosome regions (eg, 17q23) that can lead to clubfoot are detected using a fetal chromosomal microarray (CMA) genetic study.

It is the role of the obstetrician to offer additional testing, including genetic testing, and support the results of these tests with counseling. This counseling is provided by both perinatologists, also known as maternal-fetal medicine specialists (a subspecialty of obstetrics), and/or by genetic counselors. Though some families may choose not to pursue additional testing, it is the recommendation of the Society of Maternal-Fetal Medicine that it be offered [12]. A variety of tests to offer and related information to provide is listed below.

Additional imaging studies

If clubfoot is detected on ultrasound, sonographers will try to identify any additional abnormalities. Particular attention is placed on lower extremity joint motion as well as overall movement of the fetus. A fetal echocardiogram and neurosonogram are performed to ensure that there are no abnormalities in the heart or brain [12]. Once structural abnormalities of the fetus are noted, the intrauterine structures and environment are examined. The position of the fetus is essential. Additionally, any compressive structures like fibroids, amniotic bands, or synechiae are identified. Polyhydramnios or oligohydramnios can also be indicative of issues with organ systems (eg, nervous or renal).

Magnetic resonance imaging (MRI) has been shown to be useful for unknown and/or complex cases of clubfoot and is ordered at the discretion of the physician but does not aid in the diagnosis of clubfoot [13]. The sensitivity and specificity of MRI in prenatal detection of clubfoot has been reported as 100% and 85.2%, respectively. Fetal MRI is generally useful in clarifying abnormalities in addition to clubfoot when there is a concern for complex clubfoot (ie, associated with other musculoskeletal or neurologic abnormalities). Additionally, MRI has the benefit of diagnosing uterine and placental anomalies as well as characterizing the brain (ie, characterizing additional aqueduct stenosis in the case of ultrasound-diagnosed Chiari II malformation with ventriculomegaly), gastrointestinal, or other organ abnormalities that are hard to image by ultrasound. However, MRI provides no additional information about the clubfoot, and in cases of isolated clubfoot diagnosed on ultrasound, fetal MRI typically confers no additional diagnostic benefit. Considering the cost and variable availability, it is not recommended in these cases.

Fetal genetic studies

Amniocentesis and chorionic villus sampling (CVS) are generally indicated for advanced maternal age (age >35 years), abnormal biochemical screening markers (maternal alpha-fetoprotein, human

Table 1
Genetic syndromes and mutations associated with "obstetrical" complex clubfoot.

	Syndrome	Known Genes
Genetic Syndromes*	*Distal Arthrogryposes (DA) types*	
	DA1 (Classic DA)	TNNI2, TPM2, MYBPC1, MYH3
	DA2A (Freeman-Sheldon syndrome)	MYH3
	DA2B (Sheldon-Hall syndrome)	TNNI2, TPM2, MYBPC1, MYH3
	DA3 (Gordon syndrome)	PIEZO2
	DA5 (DA with ophthalmoplegia, ptosis, and retinal involvement)	PIEZO2, ECEL1
	DA7 (Trismus-pseudocamptodactyly syndrome)	MYH8
	DA8 (Autosomal dominant multiple pterygium syndrome)	MYH3
	DA9 (Congenital contractural arachnodactyly/Beals syndrome)	FBN2
	Autosomal dominant Larsen syndrome, recessive spondylocarpotarsal syndrome	FLNB
	Barth syndrome	TAZ
	Bruck syndrome	PLOD2, FKBP10
	Carey-Fineman-Ziter syndrome	MYMK
	Catel-Manzke syndrome	TGDS
	Charcot-Marie-Tooth disease type 4D	NDRG1
	Diastrophic dysplasia	SLC26A2
	Ehlers-Danlos syndrome, musculocontractural type 1	CHST14
	Ehlers-Danlos syndrome, musculocontractural type 2	DSE
	Ehlers-Danlos syndrome, vascular type	COL3A1
	Epileptic Encephalopathy	AARS
	Joubert syndrome	ATXN10, TCTN2
	Loeys-Dietz syndrome	TGFBR1, TGFBR2, SMAD3, TGFB2, TGFB3
	Marfan syndrome	FBN1, TGFBR, TGFBR1, TGFBR2, SMAD3, TGFB2, SKI
	Mobius syndrome	PLXND1, REV3L
	Multiple epiphyseal dysplasia	COL9A1, COL9A2, COL9A3, COMP, MATN3, SLC26A2
	Multiple synostosis syndrome	GDF5
	Peroxisome biogenesis disorder 7A	PEX26
	Recessive axonal Charcot-Marie-Tooth disease	LMNA, GDAP1
	Recessive Larsen syndrome, humero-spinal dysostosis, Spondyloepiphyseal dysplasia	CHST3
	Richieri-Costa-Pereira syndrome	EIF4A3
	Santos syndrome	WNT7A
	Saul-Wilson syndrome	COG4
	Shpritzen-Goldberg syndrome	SKI
	TARP syndrome	RBM20
	Van maldergem syndrome 2	DCHS1, FAT4
Other mutations*	**Affected domain**	**Known genes**
	Transforming growth factor beta (TGF-β) signaling	TGFBR1, TGFBR2, SMAD3, TGFB2, TGFB3, SKI, GDF5
	Extracellular matrix (ECM)	COL9A1, COL9A2, COL9A3, COMP, MATN3, SLC26A2, FBN1, COL3A1
	Peroxisomal defects	GDAP1, PEX26
	Proteoglycans	CHST14, DSE, COG4, TGDS

* *Genetic syndromes and other mutations as described by Sadler et al. (2019).*

chorionic gonadotropin, unconjugated estriol) in 1st or 2nd trimester, or a family history of chromosomal abnormality. Both procedures require inserting a needle into the uterus to extract amniotic fluid and placental tissue, respectively. Conventional karyotypes, fluorescence in situ hybridization, or chromosomal microarray analysis can then be completed on these cells to detect any aberrations in the chromosomes. The combination of these tests provides much information about syndromic abnormalities. Any positive results are communicated to families with the support of a genetic counselor to properly address all the sensitive and nuanced implications that are associated with genetic diagnoses.

Cell-free DNA screening

Cell-free DNA screening (also offered when maternal age is over 35 years), which does not enable microarray, may be done as early as 10 weeks' gestation when parents do not want invasive genetic testing such

as amniocentesis and CVS. This screening test uses the mother's blood sample to detect fetal DNA circulating in the maternal bloodstream and has an 83% positive predictive value to detect a limited number of chromosomal abnormalities such as aneuploidies (eg, Trisomy 21). Typically, cell-free DNA screening is only offered to patients who have a high risk of aneuploidies.

Single gene associations

Some families may want to ask whether there is a genetic component to the diagnosis of nonteratologic clubfoot. Gene associations in congenital clubfoot are starting to be scrutinized. Studies range from family studies to large database studies. Postnatally, there are no clear guidelines for referral to a genetic counselor, although a referral may be considered in cases of teratologic clubfeet or presentation with other anomalies. Prenatally, parents may be interested in understanding these genetic associations to be more informed on treatment plans and improve screening for other illnesses. Although there are no established guidelines or indications for genetic screening in isolated nonsyndromic clubfoot, referrals to genetic counseling may be considered when there is a strong family history of clubfoot or clinical features lead to suspicion of a syndromic clubfoot. There may be associated anomalies that may not be detected by ultrasound in each case, so it makes sense to offer it to everyone.

When an anomaly is detected prenatally, families should be offered genetic counseling. This provides an opportunity for parents to comprehend the possible associated syndromes and options for more testing. Often this is coordinated by a perinatologist who oversees the scans and procedures. The Society for Maternal-Fetal Medicine recommends offering genetic testing (amniocentesis vs. CVS) when a clubfoot is diagnosed [12]. In these instances, the perinatologists will automatically refer the family to a genetic counselor. Both perinatologists and genetic counselors may provide a diagnosis, administer initial counseling, and make recommendations for additional testing. They also recommend consulting with a pediatric orthopaedic surgeon to learn about clubfoot and its treatment, specifically. Though a low rate of pathogenetic causes may be identified via genetic testing, genetic counseling can help identify at-risk families, provides accurate information regarding recurrence risks, and offer access to genetic testing as appropriate. It should be noted that while there is a strong genetic basis for clubfoot, it is unlikely due to a single causative gene variant, but rather due to multifactorial or polygenic factors. Identification of genetic markers associated with clubfoot in the future should help improve screening and prevention plans and pave the way for the development of new treatments [14].

Fetal genetic studies performed in conjunction with cell-free DNA testing, amniocentesis, or CVS are part of routine prenatal care and are covered by commercial and government insurance (ie, Medicaid plans). In instances where there is concern about associated or additional anomalies in the setting of clubfoot postnatally, we recommend referral to a medical geneticist to guide testing. Genetic testing should always be performed by a qualified medical geneticist or genetic counselor since they are best qualified to discuss positive results with families. These findings can have far-reaching implications beyond just the care of the child with clubfoot and often raise sensitive issues, the discussion of which is within the scope of training of these providers. Medical geneticists and genetic counselors also play a role in helping families navigate the confusing insurance landscape. Coverage for these tests can be highly variable, and medical geneticists are best positioned to decide which tests are appropriate and obtain authorization when needed.

Regarding genetic pathways of relevance to clubfoot, the PITX1-TBX4 transcriptional pathway has been investigated as it is involved in early limb development [15]. In a series of patients with clubfoot, all of whom had a family history of the condition, characterized by a first-degree relative who also had clubfoot, 5% of those individuals had a variant in PITX1 [16]. A single missense mutation in this gene was first described in a genome-wide linkage study in a five-generation clubfoot family with nine affected individuals [15]. However, individuals in this family also demonstrated other lower limb malformations. This suggests that PITX1 (and potentially the sonic hedgehog [SHH gene]) may be responsible for syndromic clubfoot presentations [15,17]. In a larger series of 40 familial cases, PITX1 was identified as a marker for isolated clubfoot based on a specific microdeletion [18]. These findings were supported in a mouse-model study [19]. In contrast, the role of TBX4 (T-box transcription factor 4) alone in the etiology of isolated clubfoot remains to be fully understood. Instead, it has been associated with variable phenotypes (clubfoot or short, wide feet) and/or syndromic clubfoot presentations along with hip dysplasia, polydactyly, and short stature [18–20].

Mutations in RBM10 (RNA binding motif protein 10) have been associated with an X-linked, syndromic clubfoot presentation, TARP syndrome, in mouse models [17,21]. TARP syndrome is characterized by talipes equinovarus, atrial septal defect, Robin sequence (micrognathia, glossoptosis, cleft palate), and persistent left superior vena cava [17].

Genes involved in the HOX (homeobox) pathway and CASP (cysteinyl aspartate protease) pathway have been identified in clubfoot development. Ester et al. identified seven genes in the mitochondrial-mediated apoptotic genes that were associated with clubfoot phenotypes [22]. These genes are implicated in the 2q31-33 deletion region, which overlaps with the homeobox cluster D (HOXD) as well as the insulin like growth factor gene (IGFBP3) [17,23,24]. The HOX pathway is involved in limb and muscle pattern development; therefore, it is plausible that clubfoot phenotypes could develop from iteration in these genes. Finally, the filamin (FLNNB) gene pathway has also been associated with clubfoot; a recurrent FLNB E1792 deletion was identified in 0.43% of 1157 isolated patients with clubfoot in one retrospective study [25,26].

Orthopaedic prenatal counseling for clubfoot

A prenatal consultation for clubfoot with a pediatric orthopaedist may be completed in-person or by telehealth visit. Due to the abundance of internet resources available on clubfoot, parents usually have a general idea of what a clubfoot is and how it is treated.

Clubfoot is a clinical diagnosis confirmed at birth, and the pediatric orthopaedic surgeon will provide a clinical description of the lower extremity. It is often beneficial to show families a model of a clubfoot to help them understand the deformity. The baby's foot will be shaped like a kidney bean with the mid and forefoot inwardly rotated and the sole facing the groin. Parents may also ask about the need for additional testing. Most often, a thorough head-to-toe physical examination is all that is necessary. Though rarely utilized, foot radiographs, which include a dorsiflexion lateral (Turco) view and an anteroposterior view may provide additional information about hindfoot alignment if needed at any time during Ponseti treatment [27]. These are not necessary to make the diagnosis of clubfoot, but there are scenarios where they can be useful, such as determining the need for tenotomy in borderline cases [28].

Prenatal counseling should also include discussion regarding the possible association between clubfoot and hip dysplasia. There is literature to support both the presence and the absence of an association [29–33]. Consequently, the need for routine ultrasound screening is controversial. All infants should have a careful hip examination at the initial visit. The authors do not routinely obtain hip ultrasounds on babies with clubfoot unless there are other risk factors. If the infant was breech in utero, management should follow standard guidelines, which call for a hip ultrasound at four to six weeks of age in the case of a normal, stable hip examination. Additional indications for ultrasound will be at the discretion of the treating orthopaedic surgeon.

Next, causes of clubfoot are reviewed. It is best to start by explaining that the exact etiology of clubfoot is unknown and is likely multifactorial. In prenatal counseling, it is appropriate to inform mothers that an amniocentesis or CVS with microarray analysis may be performed by their obstetrician, although most parents come to their prenatal orthopaedic appointment with the results from these tests.

The most impactful element of the prenatal consultation is reassuring parents that the Ponseti method is successful and normal foot function is expected. Casting rarely occurs in the hospital. Most babies in the authors' practice are treated in the outpatient clinic within the first two weeks after discharge from the maternity ward. However, casting does not need to be started immediately after birth. This standard practice is especially comforting for families who deliver at a great distance from centers where clubfoot treatment is available. If an extended stay at the hospital for other health issues is required, the infant will begin Ponseti treatment when medically stable to tolerate the casting process. Parents are reassured that there are no issues with initiation of cast treatment several weeks later, with no impact on outcome [34,35].

If the baby has other health conditions that require urgent treatment, the pediatric orthopaedic surgeon will work with the pediatrician and other specialists to coordinate care. These issues commonly delay hospital discharge but rarely delay treatment. In the authors' experience, when cardiac, gastrointestinal, and genito-ureteral anomalies that require surgery are present, we often postpone clubfoot treatment until these procedures have been completed. The decision whether to initiate Ponseti treatment pre- or post-operatively must be individualized and based upon the timing of the other procedures. If the foot abduction orthosis cannot be used immediately after surgery on another organ system, a recurrence may occur, resulting in additional casting and possibly a repeat Achilles tenotomy. In this instance, it is prudent to delay the initiation of clubfoot treatment.

Expectations for the first appointment and casting are discussed. Recommended items to bring to the outpatient clinic/cast room are a pacifier to dip into dextrose solution (to help calm the baby through the

Table 2
Talking points at the orthopaedic prenatal clubfoot consult.

Question(s) by family	Answer(s) by orthopaedic surgeon
What causes clubfoot?	The causes are multifactorial. • Environmental factors (parental tobacco use, maternal obesity and diabetes, lower parity, and early amniocentesis). • Sex (2:1, assigned male at birth: assigned female at birth). • Genes integral to fetal limb bud development (PITX1, TBX4, HOX).
What does a clubfoot look like?	The foot will be turned inward and pointed downward. Many describe it as being shaped like a kidney bean, inwardly rotated and with the sole facing the groin.
How is the diagnosis confirmed at birth?	A careful physical examination is the best way to confirm a clubfoot diagnosis. Sometimes the prenatal ultrasound diagnosis is inaccurate.
Are there any additional tests needed after birth?	Aside from a thorough physical examination, if the baby shows no other abnormalities or neurologic findings, no other testing is needed. • Some pediatric orthopaedists routinely screen babies with clubfoot for hip dysplasia using an ultrasound. • Occasionally, x-rays of the foot help guide treatment.
What will the baby's cosmetic and functional outcome be after successful treatment?	The Ponseti method is immensely successful and normal foot function is expected in almost all cases. There are professional athletes and Olympians who were born with clubfoot. Though function is not typically affected, … • Sometimes a unilateral clubfoot may be shorter than the unaffected foot. • The calves on the side of a unilateral clubfoot may be smaller. • The ankle motion of an individual treated for unilateral clubfoot is typically slightly less the unaffected side, but not usually noticed by the individual. • Occasionally, there can be a leg length discrepancy.
What will happen on the first visit?	Most initial visits start with: • Thorough physical examination. • A brief review of the Ponseti method (as discussed in the prenatal visit, if the family had one). • Answer any other questions that arise. • Manipulation and casting.
Does the manipulation and casting hurt?	Reassurance is provided that some babies will be agitated during casting, and we suspect that there may be brief, mild discomfort, especially in the later casts in the process. We believe it is similar to that which one would feel with intensive stretching before or after exercise.
Why perform an Achilles tenotomy?	The Achilles tenotomy is performed to address the last deformity in clubfoot, equinus. This final correction in Ponseti treatment expeditiously improves the mobility of the ankle and prevents potential iatrogenic deformities such as a rocker bottom foot or flat top talus.
Where is the tenotomy performed and what type of analgesia/anesthesia will be used?	Achilles tenotomies routinely are performed under local anesthesia in an outpatient clinic, cast room, or operating room. The location is often at the discretion of the surgeon and, sometimes, institutional policy.
What is the role of bracing after cast treatment?	The foot abduction orthosis maintains the correction, preventing clubfoot recurrence and it is essential for a successful outcome.
What are the considerations and expectations at initiation of bracing?	It is important to prepare parents for the possibility that the first few days of bracing can be uncomfortable. This is likely due to the stiffness from casting and the sensitivity of the skin. This discomfort resolves promptly, typically within days, but occasionally can take up to two weeks.
Will the bracing affect my child's motor development?	There is no evidence that bracing affects motor development. Parents are often concerned that wearing the bar will interfere with the onset of walking, but by the time babies are ready to pull to stand they have plenty of daytime hours out of the brace.
What is the risk of recurrence/relapse and what is the treatment?	The risk of recurrence, identified in multiple studies, is between 31% and 56% overall. Recurrences are treated with one or more of the following: • Additional casting • Achilles lengthening • Tibialis anterior tendon transfer
What does follow-up surveillance look like?	After the foot abduction orthosis is applied post final cast removal … • The baby is seen 1 week later. • Post 1-week follow-up, babies come in monthly for 3 months while they are wearing the brace full-time. • Then they come every 6 months until 5 years old when they stop brace treatment. • After 5 years, they present every 2–3 years until skeletal maturity to ensure there are no recurrences.

gentle manipulation and cast application) and a bottle of formula or breast milk (if parents are amenable to bottle-feeding) [36,37]. It is also recommended to bring a change of clothes, including extra diapers because the casting process is frequently messy. The details of manipulation and casting are then reviewed, which include how long it takes to apply casts and the essential role parents play to comfort their children. Reassurance is provided that some babies will be agitated during casting, but it is not believed to be a painful process. Next, emphasis is placed on the importance of parental monitoring of the feet between casting visits, specifically the assessment of toe perfusion and swelling as well as evidence of cast slippage. It is helpful for the parent to take a photograph of the toe-end of the cast immediately after application to use as a frame of reference if a concern for cast slippage occurs between casting visits. If a baby remains fussier than usually and cannot be sated with feeding, diaper change, or general soothing techniques, and/or there is a problem with the cast, techniques for removal of plaster or semiflexible fiberglass casts (based upon the treating clinician's practice) are reviewed [38]. Plaster casts can be soaked off by placing the infant in a warm bath with a small amount of vinegar for approximately 1 h, and semiflexible fiberglass casts can be unraveled. Even if it is the clinician's practice to remove the cast at the appointment, families should know how to remove casts in case of an emergency.

Specifics of the Achilles tenotomy and the need to keep the infant in the same cast for three weeks until the tendon remodels are reviewed are next. This procedure is performed in 90%–95% of infants prior to the last cast application. It is important to highlight during counseling that the Achilles tenotomy is a routine part of the Ponseti method, not a failure of conservative treatment. Families should also be informed that Achilles tenotomies are routinely performed under local anesthesia in an outpatient clinic, cast room, or operating room. The procedure is well tolerated by patients and rarely requires more than a dose or two of acetaminophen, if any, postprocedure [39].

To manage parental expectations, a conversation regarding the appearance of the foot after removal of the final cast is useful. Typically, some swelling is present. Moreover, the medial malleolus is usually prominent due to the maximal abduction in the final cast, which contributes to the appearance of being "overcorrected." Explaining the reason for this and the concept of maximally abducting the foot to minimize residual deformity helps parents prepare for when the final cast is removed and bracing is initiated. A foot abduction orthosis should be available for families to see and hold. This helps them understand the brace's components and its purpose to prevent recurrences. It is important to prepare parents for the possibility that the first few days of bracing can be uncomfortable. This is likely due to the stiffness from casting and the sensitivity of the skin. This discomfort resolves promptly, typically within days, but occasionally can take up to two weeks.

Discussion of other issues related to bracing is another meaningful part of the consultation. Parents may be concerned about the impact on developmental milestones, like rolling over or sitting, but they can be reassured that there is no evidence that bracing hampers motor development [40]. Also, by the time babies are ready to roll, sit, and stand, they have plenty of daytime hours out of the brace. This time is also an opportunity to lay out strategies for successful bracing and optimal brace tolerance. From an early age, babies need to wear the brace before sleep to become accustomed to wearing it while falling asleep. Dr. Richard Ferber's previous studies on infant sleep patterns have shown that babies fall asleep and wake up many times throughout the night. Therefore, external factors and environment at onset of sleep are recommended to be the same throughout the night [41]. Consequently, it is important to encourage parents to have the patient wear the brace every day without exception and to regularly apply the brace before the child falls asleep. Clubfoot does not necessarily recur in one night, but one night out of the brace may allow infants to experience the freedom from bracing prematurely and introduce a preference for not wearing it. However, an atypical night out of the brace (eg, overnight flight or hospitalized patient) will not typically be associated with normal sleep. Hence, is not likely to lead to brace intolerance. An alliteration often used at our institution to remind parents of the nighttime routine with bracing is "bath, boots, books, and bed."

The risk of recurrence, identified in multiple studies to be between 31% and 56% overall, cannot be overemphasized. Educating parents about the influence of long-term bracing on decreasing this risk of recurrence will help them commit to the challenge of nightly bracing and strictly follow the protocol. Furthermore, reassurance can be offered that recurrences can be successfully managed, sometimes with additional casting and sometimes with an Achilles lengthening and/or tibialis anterior tendon transfer. Ongoing monitoring during the growing years, during and after completion of bracing, is an important component of clubfoot treatment and allows for early recognition of recurrences to optimize outcomes.

A detailed list of talking points between families and the orthopaedic surgeon is provided in Table 2.

Summary

Prenatal clubfoot diagnosis can be detected on prenatal ultrasound as early as 13–16 weeks of gestation. Additional testing, including amniocentesis or chorionic villus sampling with chromosomal microarray analysis may be recommended and can provide parents with detailed information about clubfoot and other health issues in their unborn child. Recent fetal cytogenetic studies have identified numerous genes involved in limb development that may predispose to clubfoot development. Prenatal testing and consultations help to provide families with detailed information about their expectant child and, more importantly, prepare all parents of children with clubfoot to have the most successful and least stressful clubfoot treatment possible.

Additional links

- Lane Wyrick, YouTube, 2021: Ponseti Clubfoot Treatment Worldwide – Feature Length Documentary
- JBJS Media, YouTube, 2017: Casting with Clubfoot Using the Ponseti Method
- EJMG, Volume 61, Issue 2: Genetics of clubfoot; recent progress and future perspectives

Consent for publication

The author(s) declare that no patient consent was necessary as no images or identifying information are included in the article.

Author contributions

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Declaration of competing interests

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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