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Tarsal Coalitions: A Review and Assessment of the Incidence in the Amish Population

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Objective: *The incidence of tarsal coalitions in the general population is not clear; it is supported that there is a genetic component to the congenital form of tarsal coalitions. Although, there have been no known studies on the Amish, it is believed that the Amish have a greater percentage of coalitions due to their restricted gene pool. This study was performed to show that there is an increase of tarsal coalitions in the Amish community.*
Materials and Methods: *This was a 10-year study of 33 Amish patients with tarsal coalitions involving the talo-calcaneal, calcaneo-navicular, and talo-navicular coalitions. A total of 38 cases were reviewed.*
Results: *Of the 38 total cases reviewed, 29 of the 38 involved the talo-calcaneal joint, a 76.3% incidence of the talo-calcaneal coalition. The calcaneo-navicular joint was involved in 8 of the 38 cases, or a 21% incidence. Only 1 patient, a 2.6% incidence, was reported to have a talo-navicular coalition. More males were affected with coalitions with overall male to female ratio of 18:11 respectively. There were several bilateral cases noted, with more calcaneo-navicular cases noted to be bilateral than any other type of coalition. It was noted to be a 25% incidence.*
Discussion: *The results in this study suggest that tarsal coalitions are indeed inherited, likely as a multi-factorial disorder of autosomal dominant inheritance. From these results, tarsal coalitions are not as rare of a phenomenon as previously believed especially in the Amish community.*

Key Words: Coalitions, Tarsal bones.

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The Amish are a Christian denomination of people that pursue an agriculturally based, "simple" lifestyle guided by faith. There are several sub groups, or "Orders", of Amish that exist within the U.S. These religious orders not only dictate how the Amish are to conduct their lives, but they also determine how the groups differ.

What is common among the different orders is the attempt to preserve elements of culture consistent with rural living of earlier centuries. Accordingly, the Amish, to varying degrees, isolate themselves from contemporary America. Economics, in large part, makes this a necessity as farming and the supporting trades are traditionally their primary source of revenue and sustenance. Therefore, communities are based in the country, not the city.

Settlement away from larger urban centers also serves to decrease the likelihood of mingling with the non-Amish, referred to as "the English", and facilitates their rejection of modern life conveniences including motor vehicles, electricity and related implements.

There are approximately 150,000 Amish in America. They are descended from German-Swiss immigrants. Two major migrations of the Amish to America are noted to have taken place with the first in the mid 18th century, and the second in the mid 19th century. The migrations were small in number with the first totaling around 200 individuals.

Religious practices and lifestyle pursuits mandate current day, self sanctioned isolation of the Amish people. The original American ancestors were few, and marriage within the group has predominated through the years. The result is a limited gene pool and a greater incidence of inherited disorders perhaps a result of the Founder Effect, which is a form of "genetic drift". Genetic traits that would otherwise be uncommon in the gene pool of the overall population may present with concerning frequency in a small adjunct of that pool. The Amish of Geauga County Ohio may serve to illustrate this. They comprise roughly ten percent of the county's population, yet make up half of the county's documented special needs cases.

The Amish believe in folk medicine: faith healing and herbal treatments are common practice and modern medicine is avoided. In fact, the Amish distribute self written and published guidelines passed down through the generations for treatment of ailments ranging from fever to burns. By virtue of their belief that ultimately it is God that heals; the Amish are reluctant consumers of modern medicine. Should an individual seek medical treatment, the indication is that he has exhausted all traditional recourse or it is an emergency.

Spanning 10 years, from 1998 to 2007, twenty-five Amish patients presented with similar complaints of foot and ankle pain. Upon clinical and radiographic examination, they were diagnosed with tarsal coalitions.

The author practices in Geauga County, Ohio, which is the world's fourth largest Amish settlement. He has had the privilege of treating the Amish community and has treated them for a variety of conditions but has noted an increased incidence of tarsal coalitions, a relatively rare condition, in the Amish community.

Tarsal coalition is an abnormal cartilaginous, fibrous or bony, union between two or more bones of the midfoot or rearfoot. This union decreases, or restricts, necessary motion of the bones involved. Tarsal coalitions can produce a remarkable symptom complex that may ultimately be identified as rigid peroneal spastic flatfoot. [6] An understanding of tarsal coalitions and the associated symptom complex will facilitate proper diagnosis and treatment.

Tarsal coalitions may be classified in several ways. These classification systems help to describe and provide information relevant to the development of a surgical treatment plan thereby increasing the likelihood of a positive outcome.

A tarsal coalition may be classified based on the type of tissue forming the union as follows: a synchondrosis (cartilaginous union), a syndesmosis (fibrous union), a synostosis (osseous union) or a combination.[6] Coalitions can also be classified according to their anatomic constituents. Tachdjian[7] provided a classification that subdivides coalitions into the bones that are abnormally united, or that are part of a complex malformation deformity. This classification is simply a descriptive one, but it does suggest the importance of assessing other areas of the foot as well as the rest of the body when a local or isolated coalition is identified.[6] It was not until Downey proposed the articular classification system that the basis for communication regarding the surgical treatment was made possible. The classification system includes the following criteria: the age of the patient, the type of coalition, and the degree of secondary arthritic changes. The conditions are then further subdivided into the type of coalition and whether they are extra-articular or intra-articular. [6] This system assumes that these are the most important criteria in determining proper treatment plans for the patient.

Tarsal coalitions are not a modern phenomenon. They have been identified and recorded for several hundreds of years. Archaeological findings have demonstrated this anomaly in a Mayan civilization in Guatemala[8,9] and in a pre-Columbian Indian civilization in Ohio[10], both dated circa 900- 950 A.D. Buffon was probably the first to recognize the occurrence of tarsal coalitions with a written description in 1769.[11] An updated version exists in the Museum of the Royal College of Surgeons of England, described by John Hunter probably dating back to 1760 – 1770.[8] Sir Robert Jones made the first detailed clinical record of peroneal spastic flatfoot available in 1897.[12]

However, he did not correlate it with tarsal coalitions. It was not until the work of Slomann in 1921[29], Badgeley in 1927[30] and Harris and Beath in 1948[31] that the correlation between tarsal coalitions and peroneal spastic flatfoot was made.

Prior to the discovery of radiography, clinicians had to rely on clinical examination alone to diagnose a tarsal coalition. Many years passed between the earliest anatomic descriptions and the development of radiography, and many additional years passed before identification of the relationship between tarsal coalitions and peroneal spastic flatfoot. Advanced imaging techniques have simplified the diagnosis of tarsal coalitions today. Although, much has been written about tarsal coalitions, little is known about the exact inheritance potential in certain cultures. The medical community remains uncertain about the cause of coalition in many respects. What is understood with certainty is that there seems to be a number of causes and they may be congenital or acquired.

Congenital coalition is more commonly recognized and reported, although the mechanism of congenital coalition is not clear. Pfitzner[13] suggested that tarsal coalitions were formed by the incorporation of accessory ossicles into adjacent tarsal bones. Many supported this theory as one possible cause, until Harris, in 1955, disproved it as the sole cause by demonstrating a tarsal coalition in a fetus.[14]

These results confirmed the theory proposed by Leboucq[15] suggesting that congenital coalition results from the failure of differentiation and segmentation of primitive mesenchymal defect. This theory links congenital coalitions to a heritable defect or to an insult in the first trimester of pregnancy. Since then, many authors have suggested a hereditary component of coalitions. Recent field studies support evidence for a mutation during formation which is passed onto future generations as an autosomal dominant gene.[6,16] Recent histopathologic evidence has been found to further support Leboucq's theory.[17] Accordingly, the generally accepted theory is that congenital coalitions are caused by a genetic mutation to an autosomal gene that results in the failure of differentiation and segmentation of primitive mesenchyme.[5,6]

The coexistence of other abnormalities with coalitions provides more evidence of an inherited component. Symphalangism, a well documented skeletal syndrome of ankylosis phalanges, demonstrates an association with coalitions. Several reports of multiple generations of individual families exhibiting both deformities exist within medical literature.[18-23] Other less common examples of abnormalities noted to coexist with coalitions include various major limb anomalies. There have been reports of massive coalitions linked to syndromes of greater complexity such as Nievergelts-Pearlman syndrome, Aperts syndrome and Osebold-Remondini syndrome.

Acquired coalitions must also be mentioned. Several reports suggest that a coalition may result from arthritis, infection, trauma, or neoplasm.[24] It is noted that these causes are less common in pediatric and adolescent patients.[25] However, when all age groups are considered, acquired coalitions are common and are a frequent cause of peroneal spastic flatfoot.

This paper examines the relationship between the Amish population and the congenital tarsal coalitions. Coalition cases with coexisting anomalies were excluded from this report. Although it exceeds the scope of this paper, it should be noted that a relationship does exist between associated anomalies and tarsal coalitions

Figures 1 and 2 above demonstrate a male patient prior to surgery demonstrating his inability to invert as compared to the contralateral foot.

Materials and Methods

The incidence of tarsal coalitions in the general population is not known with certainty. Various authors have attempted to show the frequency (Table 1), but these studies have been based on selected groups and do not identify an accurate incidence within the general population.

A survey of relevant literature suggests a tarsal coalition incidence within the general population of 1% to 2%. However, one could argue that respective cohorts of these studies were not representative with regard to age or sex. Some have suggested a greater incidence of tarsal coalitions in males. Two studies support this, citing the following male to female ratios: 12:5 (26) and 4:1 (27). Therefore, the actual incidence in the general population may be higher or lower.

Reports demonstrate little proclivity in the occurrence of coalitions along racial lines.[5] It is estimated that people of Asian, African, and Latino descent have a slightly higher incidence than most. There have been no studies comparing the incidence of the Amish community to date.

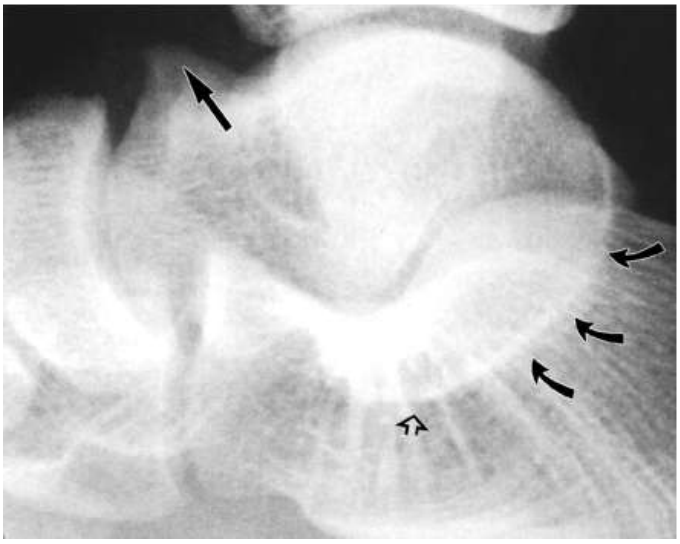

Figure 3 Lateral radiograph used to evaluate a tarsal coalition of a 25-year-old male. Talar beaking is demonstrated (solid straight arrow) as an upwards flaring of anterosuperior aspect of talar head. The C sign as demonstrated by the curved arrows, which is continuous cortical contour extending from medial aspect of talus to sustentaculum tali. The middle subtalar facet is not seen on this view. Note that sustentaculum tali shown as the open arrows, has a curved undersurface instead of a normal flat contour. The talar neck appears shorter than normal.

The authors of this paper studied 33 Amish patients from Middlefield, Ohio, located in Geauga Country, over a ten year period from 1998 to 2007. A total of 42 coalitions were diagnosed, however, only 38 cases were included in this study. Ethical approval and informed consent was obtained for this study The patient's ages ranged from 7 to 54 with a median age of 15 years. The male to female ratio was 21:12 respectively.

Of the 38 patients in this study, four demonstrated bilateral coalitions. (Table 2)

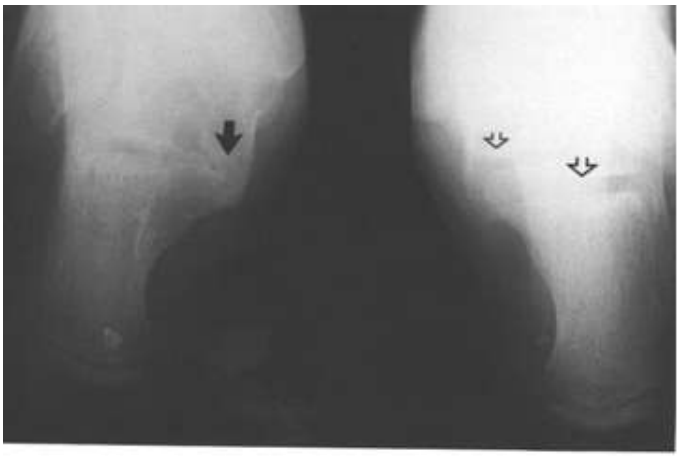

Figure 4 A calcaneal axial radiograph taken of both feet demonstrating the middle and posterior facet as a comparison.

Each coalition was broken down into the respective coalition and further subdivided into the affected foot as shown in Table 3.

Each of the 33 patients that presented to the office had a chief complaint of pain. They related a deep, aching pain that increased with prolonged activity. The pain was reported to decrease with rest. Each patient was able to localize the pain to the appropriate tarsal area. Activities noted to increase their pain were walking, especially over uneven terrain, long periods of standing, and athletics.

Limited range of motion of the subtalar joint was the second most common complaint. Patients complaining of limited motion demonstrated a pronounced decrease in subtalar joint inversion. The total ROM required at the subtalar joint in a normal gait is 8 to 12 degrees of frontal plane motion. An average minimum ROM of 4 to 6 degrees of inversion of the calcaneus is needed with supination of the STJ. Also, 4 to 6 degrees of eversion of the calcaneus with subtalar joint pronation is also needed for ambulation. Of the 33 patients analyzed, roughly half had an average of 2 degrees of inversion noted on physical examination with the patient standing (Figures 1 and 2).

With the patient sitting on the examination table, they were asked to dangle both legs over the side placing them in a dependent position. A normal foot in a dangling, dependent position will demonstrate equinus, mild rearfoot varus, and slight forefoot supination with a well-formed longitudinal arch. The patients of this study exhibited otherwise. With their legs in a dependent position over the side of the exam table, each patient revealed a foot that failed to adopt the normal equinus position. Additionally, the foot demonstrated a neutral to valgus position, with no supination of the forefoot.

A small percentage of the patients who presented to the office complained of tonic muscle spasms, namely of the peroneal muscles. Tarsal coalitions may cause peroneal spastic flatfoot, but this is not the same condition. Electrodiagnostic studies have confirmed the absence of a true neurologic clonic spasm.[28] The peroneus brevis is usually the muscle that is the most significantly involved, as was true in the cases seen here. However, it is also possible to have spasm of the tibialis posterior, tibialis anterior and peroneus longus muscles.[5,6] It is believed that the clonic reaction of the peroneal muscles to passive inversion stress is actually a manifestation of shortening of the muscle- tendon unit caused by a fixed valgus position of the subtalar joint.[5]

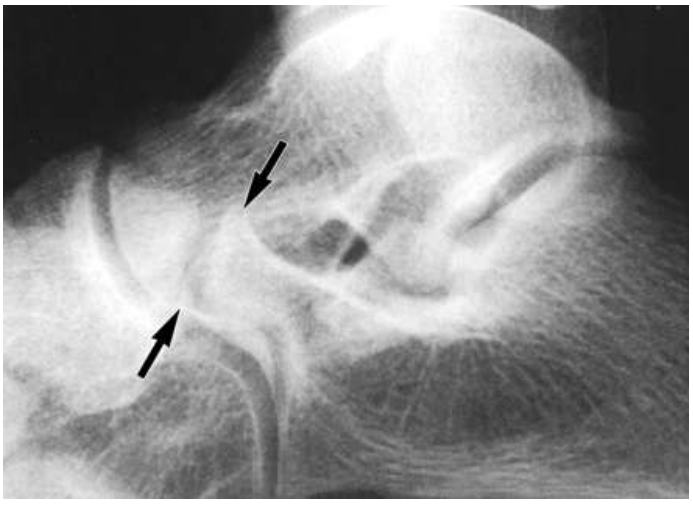

Figure 5 A lateral radiograph used to evaluate a calcaneo-navicular coalition of a 26-year-old woman. Showed above is that of the "anteater" sign. The anterior process of calcaneus (between arrows) is enlarged and elongated and has blunt tip like anteater's snout. This is one of the hallmarks in identifying the coalition.

Upon completion of the physical examination, radiographs were taken of the suspected tarsal coalition. Routine radiographs often lead to an accurate diagnosis of tarsal coalitions.[6] More advanced technologies, such as computed tomography (CT) or magnetic resonance imaging (MRI), are usually reserved for when standard radiographs are inconclusive or when identification of the coalition in more detail is required. Radiographs are the most cost-effective modality of choice.

The dorsal-plantar foot view, a standard for identifying coalitions, is the view of choice for talo-navicular coalition. The lateral foot, calcaneal axial and Harris-Beath projections serve to produce the best images for identification of talo-calcaneal coalitions. The medial oblique foot view is best reveals for the calcaneo-navicular coalition.

The lateral radiograph foot view demonstrates a talo-calcaneal coalition of the middle facet that is directly visualized, as the joint space is absent between the talus and calcaneus. This is referred to as a C sign or a halo and this, combined with the absence of the middle subtalar facet, is the hallmark of the talo-calcaneal coalition. With the absence of the STJ, there is sclerotic enhancement of the sustentaculum tali. Also, it is noted that there is flattening of the lateral talar process.

The calcaneal axial, also known as Harris-Beath view, radiograph seen above, was taken to evaluate the status of the middle and posterior facets. The above image demonstrates a normal subtalar joint with the middle and posterior facets both parallel to the ground and to one another. This image reveals an angulated middle facet that is typical of a talo-calcaneal coalition. The middle facet of the left foot, upon measurement, was noted to be angled more than 25 degrees than that of the right view. A talo-calcaneal coalition is strongly supported here.

The calcaneo-navicular coalition, demonstrated via medial oblique view, was the first coalition to be identified radiographically.[29] The coalition is thought to be best appreciated with a medial oblique view, but is also suggested on a lateral view. Both examples are demonstrated in figures 5 and 6.

A comma sign on a medial oblique view is noted as the protrusion of the calcaneus toward the navicular. This is considered one of the hallmark signs to identifying a calcaneonavicular coalition.

Most tarsal coalitions may be diagnosed by clinical examination combined with traditional radiographs. CT and MRI images offer the ability to obtain multiplanar cross-sectional images of the foot with minimal bone overlap. However, these modalities should be reserved for cases in which radiographs are inconclusive for evaluation of a coalition of the anterior facet of the subtalar joint, and for cases in which the physician desires more detailed information regarding a specific coalition. With CT scans, the feet are assessed simultaneously after proper positioning in the CT gantry. Proper assessment of tarsal coalitions requires both axial and coronal views of the ankle and foot.

Coalitions and degenerative changes are usually easier to diagnose with CT scans than with standard radiographs. This form of imaging is extremely useful for preoperative planning. CT data are usually unequivocal for osseous involvement. The CT scan does have its limitations when it comes to fibrous coalitions, however. MRI is superior in differentiating osseous from fibrous coalitions because of the superior ability to image cartilage and soft tissue. MRI has proven to be a useful, and increasingly preferred, cross-sectional imaging modality for coalitions.

When it comes to treatment, once conservative treatment had failed, surgical options were discussed with each patient and his or her family. Twenty-five patients elected the need for surgical intervention. The surgical procedure plan was specific for each patient. The patients, age, articular involvement, secondary arthritic changes and the classification of coalitions encountered on examination were taking into account during the planning of each procedure. The procedures are outlined in Table 4.

Results

Over this ten year study taken during 1998 to 2007 in the Amish population of Middlefield, Ohio, the analysis encompassed 38 cases with a total of 33 patients. Of the 38 cases reviewed, 29 of the 38 cases were found to involve a talo-calcaneal coalition, a

calculated relative incidence of 76.3%. Of these 29 cases 3 of them were found to occur bilaterally, an incidence of approximately 10%. Of the talo-calcaneal coalitions, the male to female ratio is 18:11 respectively. Sixty-two percent of the talo-calcaneal coalitions were found in male population. A 2:1 male to female ratio of talo-calcaneal bilateral coalitions was found. Each of the talo-calcaneal coalitions were found to be of the middle facet.

Of the 38 cases reviewed, 8 of the cases were reported to involve a calcaneo-navicular coalition, a calculated relative incidence of 21%. Of these 8 cases, 2 bilateral cases were found encompassing approximately 25% of the cases. The male to female ratio for the calcaneo-navicular coalitions was found to be 6:2 respectively, approximately a 75% incidence found in males. Both bilateral cases were found in the male population analyzed. Only one patient was reported to have a talo-navicular coalition, a calculated relative incidence of 2.6%. This one patient was a female and happened to be the oldest of the patients included in the study.

Patients included in the study had a wide range of age that included 7 to 54 years of age. The median age of the patients was 15 years of age. More than half of the patients were males with a male to female ratio of 21:12 respectively of the 33 total patients. It was not assessed if these patients had other family members with the same conditions.

Eight of the 33 patients reviewed did not undergo surgical intervention. These patients were just treated conservatively. Of these 8 patients, 3 were males and 5 were females, a relative incidence of 37.5. All of these cases were unilateral.

Discussion

Tarsal coalition is a condition characterized by a restriction or absence of movement of the hind or mid-foot, secondary to an abnormal union between two or more bones at this site. Coalitions may be cartilaginous, fibrous or bony and the condition may be congenital or acquired. The overall prevalence of tarsal coalitions in the general population is not clear, however, it is reported that it is about 1%.[1] Since many coalitions remain asymptomatic in life, however, the actual figure may be slightly higher. Race does not appear to be significant and, of those affected, between 50% and 60% have the condition bilaterally.[1] The most common coalitions are either between the anterior process of the calcaneus and the navicular bone or between the talus and calcaneus, usually involving the middle facet.

The median age of patients reviewed was 15 years of age. It has been found that the onset of symptoms may correlate with the beginning of ossification of the coalition. A talo-navicular coalition ossifies from the ages of 3 to 5 years, a calcaneo-navicular coalition ossifies from the ages of 8 to 12 years and a talo-calcaneal coalition ossifies from the ages of 12 to 16 years.[6] Since most of the patients in the study were found to have a talo-calcaneal coalition, the median age of 15 falls into the ossification range of 12 to 16 years, further supporting the diagnosis of the coalition.

It has been reported in numerous literature that the talo-calcaneal coalition and calcaneo-navicular coalition are by far the most common anatomic types of this condition. The current consent is that the talo-calcaneal and calcaneo-navicular coalitions account for a majority of all coalitions, with the talo-calcaneal coalition slightly more common. This was found true over the 10 years studied. The Amish population had a 76.3% affected with a talo-calcaneal coalition, in comparison to the calcaneo-navicular coalition, which made up 21%. This data supported the literature data up to date in regards to most common, presentation, occurrence, incidence, and gender most likely to be affected.

Previous studies of the closed Amish population have proven to be a valuable tool in the field of genetics. Isolated populations, with homogenous genes such as the Amish in Geauga County, Ohio, offer genetic researchers an unparalleled insight into disease and genetics. These closed populations, whether by geography or religion, were created by just a few families, called the "founder effect", and built on generations of inbreeding.

The Amish have higher rates of inherited diseases caused by bad, recessive genes that are diluted in the general population but remain captive in closed societies. This increases the odds that distant relatives, who are each carriers of rare disorders, will marry and produce afflicted children.

Although tarsal coalitions are considered fairly uncommon in the general population, it is believed through this paper that the percentage of coalitions is higher in certain cultures of the population. Literature has argued that there is a pre-disposition in certain cultures, such as Orientals, African-Americans, and the Latino communities. Although these cultures have been studied more than others, it is difficult to obtain the accurate incidence of coalitions. Thus far, over this 10-year study, it is felt that the 33 patients who did come in to seek medical attention, is a fairly large number in this small population. Keep in mind that these patients came in for medical advice because they had to.

It is difficult to tell how many other people in the Amish community have coalitions and have decided that the pain or lack of motion is tolerable. It should be noted that there were 5 other coalition cases that presented in the 10-year time span who did not want to be included in the study.

The results of this study suggest that tarsal coalitions are indeed inherited, more than likely as a multi-factorial disorder of autosomal dominant inheritance. From these results, tarsal coalitions are not as rare of a phenomenon as previously believed, especially in the Amish community.

Authors	Material	Incidence
Rankin and Baker (45)	Military personnel	0.04% (24 cases in 60,000 young adults)
Shands and Wentz (46)	Children clinic	0.9% (11 cases in 1,232 children)
Vaughan and Segal (47)	Army personnel	1.4% (28 cases in 2,000 patients)

Table 1 The incidence of coalitions as described in the literature.

Coalition Type	Gender	Age
Talocalcaneal	Female	15
Calcaneonavicular	Male	13
Calcaneonavicular	Male	7
Talocalcaneal	Male	10
Talocalcaneal	Male	15

Table 2 Bilateral coalitions (gender and age).

Coalition Type	Bilateral Feet	Right Foot	Left Foot
Talocalcaneal	3	11	12
Calcaneonavicular	2	3	1
Talonavicular	0	0	1

Table 3 Tarsal coalitions reviewed over a 10 year study in the Amish culture.

Surgical Procedure	Quantity
CN Coalition Resection	6
Triple Arthrodesis	1
STJ Resection with Fat Graft	9
STJ Fusion	4
STJ Resection with MBA Implant and Fat Graft	1
STJ Fusion with TAL	1
STJ resection	4
STJ Fusion with Calcaneal Osteotomy and Lateral Column Fusion	1
TN Fusion	1
CN Bar Resection with MBA Implant	1
STJ Resection with MBA Implant	1

Table 4 Surgical procedure performed during 10 year study.

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