Brachymetacarpia and brachymetatarsia are rare congenital presentations denoted by shortening of metacarpals and metatarsals respectively, in which the deformity usually presents around childhood/early adolescence.

The aetiology is usually congenital besides several other syndromic or endocrinologic associations.

Cosmetic issues such as absence of a normal-looking knuckle while making a fist or disruption of finger-tip curvature besides functional issues are the main indications for surgical intervention in brachymetacarpia.

In the foot, apart from cosmetic issues, pain due to transfer metatarsalgia as well as callosities along with toe deformities which lead to difficulty of using footwear are the main indications for intervention.

Lengthening of the affected bone, either acute with grafting or gradual, is the mainstay of treatment. Gradual lengthening can be either single-stage as in callotasis, or two-stage where the primary procedure is followed by bone grafting after the length has been achieved.

Adolescence, specifically between 12 and 15 years, is the preferred period for surgical intervention in these cases.

Keywords: bone lengthening; brachymetacarpia; brachymetatarsia; congenital difference; congenital malformations; distraction osteogenesis

Introduction

Brachymetacarpia and brachymetatarsia are rare congenital differences of the extremities in the paediatric and adolescent age groups. They are defined by the shortening of metacarpals and metatarsals, respectively, due to premature fusion of the growth plate as well as due to pathology in the bone itself. Brachymetacarpia is defined on the basis of the formula given by Aydinlioglu, that is, first metacarpal = 0.67 × second metacarpal = 0.71 × third metacarpal = 0.78 × fourth metacarpal = 0.84 × fifth metacarpal.1 Brachymetatarsia is defined with reference to the parabola formed by joining the metatarsophalangeal joint (MTP) of the toes.2 It is present when one or more of the metatarsals ends 5 mm proximal to this parabola. They both occur more commonly in females than in males.3 While the prevalence of brachymetacarpia is unknown, that of brachymetatarsia varies considerably in different studies between 0.002% and 0.05%.4 The third, fourth and fifth metacarpals are the most commonly involved in brachymetacarpia (Fig. 1). In the foot, affection of the first and fourth metatarsals are most common, with 72% of cases being bilateral (Fig. 2).5 The deformity usually becomes apparent by the age of around 10 years. The aetiology of the deformity can be either congenital or acquired. Although congenital cases are most commonly noted, infection and trauma are the most common causes of acquired cases. Apart from being idiopathic, numerous syndromes and endocrinopathies are associated with brachymetacarpia and brachymetatarsia but they also may be present simultaneously as a sporadic congenital entity (Table 1).4,6 Sporadic mutations have also been reported to cause this anomaly.7,8

Fig. 1 A case with brachymetacarpia of the fourth metacarpal is shown. Please note the abnormal palmar crease at the MCP4 joint (arrow).
anomalies such as syndactyly, cleft hand and symbrachydactyly may also be seen in these cases whereas syndactyly, polydactyly, brachymesodactyly and accessory navicular have been reported in the foot.9–11 Deformities of the nail and nail bed can be associated in both of these conditions as well.12

A literature search was performed using PubMed, Embase, Google Scholar, and the Cochrane database in April 2020. The systematic search was carried out in accordance with the Cochrane Collaboration, Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) guidelines. The keywords used were ‘brachymetacarpia’ and ‘brachymetatarsia’. The search was also replicated using the appropriate MeSH terms. The following filters were used to obtain the probable article candidates: humans and English. No limitations were set with regard to article type, treatment strategy and number of patients included. Additional articles were identified through the ‘related articles’ feature. All bibliographies were checked for further probable studies for inclusion. Full texts were obtained after screening of titles and abstracts.

**Biomechanical considerations and surgical indications**

**Brachymetacarpia**

Impaired hand function, apart from cosmetic concerns, is the second most common complaint among patients with brachymetacarpia. The key functions of the hand are precision pinch, key pinch, chuck grip and power grasp. These functions require coordinated action between the muscles, tendons and bones in the hand. Lumbricals, interossei, thenar and hypothenar muscles and the long flexors are the main muscles with their tendinous portions being involved in hand function.13 The shortening of the metacarpal, as is seen in brachymetacarpia, can lead to relative lengthening of the corresponding muscle–tendon unit. This may result in reduced force generated from the contraction of these muscles, which in turn can manifest as weakness of hand function.

Metacarpophalangeal joints (MCP) are considered to be the most important joints in hand function because they contribute to 77% of the arc of motion of the fingers.13 Also, most prehension grips require extension and some degree of abduction at the MCP joint. The level of the MCP joint is altered in brachymetacarpia which can affect both range of motion and thus hand function. The third ray provides the most individual flexion force, so in presence of third metacarpal involvement, the grip and grasp function is definitely affected.13 Although individual forces of the ring and little finger are lesser, they are important factors in the ability to hold objects in the palm. Hence, involvement of the fourth and fifth metacarpal can also lead to weakened grasp function.

| Syndromes                      | Endocrinopathies                          |
|--------------------------------|-------------------------------------------|
| Familial                      | Neonatal hyperthyroidism                  |
| Down syndrome                 | Pseudohypoparathyroidism                  |
| Turner syndrome               | Pseudopseudohypoparathyroidism            |
| Holt Oram syndrome            | Epiphyseal dysplasia                     |
| Klenefelter syndrome          | Myositis ossificans                      |
| Larsen syndrome               | Achondroplasia                            |
| Apert syndrome                | Chondrodysplasia punctata                 |
| Multiple hereditary exostosis | Acromelic dysplasia                       |
| Nance Horan syndrome          | Albright hereditary osteodystrophy        |
| Dyschondroosteosis            |                                           |
| Cri-du-chat syndrome          |                                           |
| Poland’s anomaly              |                                           |
| Rett syndrome                 |                                           |
| Robinow’s syndrome            |                                           |

**Fig. 2** A case with bilateral brachymetatarsia of the fourth ray is shown. Due to the severe shortening an elevation and malrotation of the toes has developed.
Moreover, the thumb is considered the most important digit in humans in regard to its placement in relationship to other digits, circumduction and opposition. This function requires a concerted movement at the trapeziometacarpal as well as the MCP joint which may be altered due to a short first metacarpal. These complex motions are actively controlled by the intrinsic and extrinsic muscles and not by articular constraints alone. The muscles may not have their full power achieved due to relative lengthening in first metacarpal brachymetacarpia. These factors may eventually lead to weak pinch function if the first metacarpal is involved.

Despite the presence of sparse literature regarding the surgical procedures for addressing brachymetacarpia, there are no standard guidelines on the indications for surgery in these cases. The various indications cited in the published literature are tabulated below (Table 2).

In summary, cosmetic issues are the most common complaints of patients with brachymetacarpia which warrant a surgical intervention. In the review of the literature, cosmetic issues accounted for around 50% of the surgical interventions in cases of brachymetacarpia. The absence of a normal-looking knuckle while making a fist and the disruption of the curvature of the extended fingers are the two most common cosmetic reasons presented by the patients. The most common technique in regard to metacarpal lengthening is gradual distraction/bone lengthening with an external fixator (Fig. 3). Cosmesis as an indication is more common for the third to fifth fingers.

Hand function may also be affected when either the first or fourth/fifth metacarpal is involved. Involvement of these rays leads to disruption of pinch and grasp function. Alterations of the transverse metacarpal arch lead to deficient grasp and grip power. Patients are mostly affected by cosmesis in congenital brachymetacarpia whereas, in acquired cases, it is the functional anomaly resulting in suboptimal hand grip that forms the

### Table 2. Review of various studies with the indications for their interventions in brachymetacarpia (multiple indications may be present in the same patient)

| Studies          | Indications (number of hands) | Mean age (Range) (years) | Outcome                                                                 |
|------------------|------------------------------|--------------------------|-------------------------------------------------------------------------|
| Lam et al (2019) | Restricted motion at MCP joint (7/7) | 22.8 (16–30)            | Range of motion improved in all patients                                |
| Ho et al (2018)  | Pain at MCP joint (1/1)       | 13.0                     | Complete resolution of pain                                             |
| Dana et al (2017) | Poor grasp (8/15)             | 12.0 (9–14)              | Not mentioned                                                           |
| Bozan et al (2006) | Cosmesis (8/8)               | 18.6 (13–20)             | Improved cosmesis in all patients                                      |
| Kato et al (2002) | Cosmesis (6/6)               | 13.3 (10–19)             | Improved cosmesis in all patients                                      |
| Miyawaki et al (2002) | Poor pinch function (4/4)     | 7.3 (5–11)               | Improved pinch function in all patients                                |
| Arsalan (2001)   | Poor pinch function (4/6)     | 12.0 (10–14)             | Pinch function and cosmesis improved in all patients                   |
| Saito et al (2001) | Cosmesis (3/3)               | Not mentioned            | Cosmesis improved in all patients                                      |

**Fig. 3** An adolescent girl with brachymetacarpia of the fourth and fifth rays (A) received gradual lengthening of the fourth metacarpal using a monolateral mini-fixator (B). The same procedure was carried out on the contralateral side beforehand (C).
chief complaint. Limited motion and pain at the MCP joint are rare indications with which the patient might present. Brachymetatarsia

The primary stress during weight bearing is taken by the plantar aponeurosis (60%) and metatarsals (25%). The so-called windlass effect of the plantar aponeurosis becomes evident during toe extension when the aponeurosis stretches around the MTP joint. This is hampered if any of the metatarsals is short, therefore leading to increased stress on the metatarsals rather than the aponeurosis. Shortening of any metatarsal may also lead to stretching of the transverse metatarsal ligament, consequently leading to altered forefoot contact with the ground and excessive pressure. Additionally, in brachymetatarsia of the first metatarsal, the effectiveness of the ‘toe-off’ phase of the gait cycle is reduced, which may lead to transfer metatarsalgia. A rigid pulley system is created during toe-off phase of gait due to which the intrinsic and extrinsic muscles of the foot act inefficiently, which has been proven by electromyographic studies.

Hence, in brachymetatarsia the shortening of the metatarsal can lead to suboptimal functioning of the muscles leading to fatigue pain in the leg and foot. Studies have also reported that increased shortening of the first metatarsal leads to transfer metatarsalgia from the second through fourth metatarsals. The chronicity of symptoms may also lead to contracture of the extrinsic and intrinsic muscles.

The toes can be affected secondarily to brachymetatarsia which warrants surgical intervention. Due to the shortened metatarsal and subsequent stretching of the transverse metatarsal ligament, the adjacent toes are pulled towards the affected toe and the affected toe displaces dorsally leading to plantar callosity over head of the short metatarsal and dorsal callosity over the toe. Global transverse digital deviation may also occur in longstanding cases leading to windswept deformity of the toes. Hallux valgus has been associated with third and fourth metatarsal brachymetatarsia, because the first and second toes deviate towards the ‘empty space’ created by the shortened ray. We have also seen hallux valgus in brachymetatarsia of the second ray.

Secondary cavus may also develop secondary to first metatarsal brachymetatarsia which can further lead to hindfoot deformities. The metatarsal inclination angle is the angle formed by the plane of metatarsal with the floor, which is around 30 degrees in a normal foot. This is increased in brachymetatarsia because the shortened metatarsal has to incline more plantarwards to make contact with the floor. This increased inclination creates a secondary cavus deformity which might progress to involve the hindfoot.

The various indications for surgery are tabulated below (Table 3). As in brachymetacarpia, cosmesis is the most common indication for surgical intervention in brachymetatarsia. It is particularly important for young females who avoid wearing open shoes or going barefoot so as to conceal the deformity. It may consequently lead to psychological distress in the affected patients. These cosmetic deformities can be managed conservatively (e.g. insoles) if the shortening is less than 5 mm as compared to the Lelivre parabola that connects congruously the metatarsal heads on an anteroposterior radiograph of the foot. Lee et al quantified this shortening of 10 mm as compared to the adjacent metatarsal which may lead to presence of symptoms needing intervention.

Pain is judged to be the second most common indication for intervention in brachymetatarsia. It usually varies in its location in the foot. The most common site of pain is the head of adjacent metatarsal due to transfer metatarsalgia. Pain on exertion such as on prolonged walking or in athletes occurs as a diffuse pain in the forefoot. Patients may also experience pain due to altered mechanics of the foot also lead to pain on the dorsum of the foot. These are formed due to dorsiflexion of the toe in the crowded web space pushed by the two adjacent normal toes.

Various toe deformities also occur in conjunction with brachymetatarsia which may warrant surgical correction. The common associated toe deformities are hallux valgus, hallux varus and hammer toes. It should be emphasized that in the presence of secondary toe deformities, these should also be addressed in addition to the shortening. Conversely, a secondary hallux valgus should only be corrected in combination with lengthening of the second metatarsal to prevent a recurrent hallux valgus. These may occur secondary to prior surgeries on the forefoot. Patients may also experience pain due to the toe deformities rather than brachymetatarsia itself. Difficulty in using footwear can be present due to the toe deformities or painful callosities. Correction of the toe deformities along with brachymetatarsia (Fig. 4) is needed in these cases. Impaired hygiene of the web space due to the toe deformities can be present as well.

Timing of surgery and surgical options

Brachymetacarpia

Adolescence, specifically between 10 and 15 years of age, is considered to be an ideal time to intervene in affected cases. At this age, patients can fully comprehend the treatment, its course and complications. The final length to be gained is also better estimated in adolescence since growth plates are rather close to fusion.
by this time. Another facet of a good outcome is postoperative rehabilitation which is extremely important to prevent stiffness of joints, and this as well can be better managed in adolescence.\textsuperscript{20} With regard to callus distraction, the healing index is defined as the number of days required for consolidation of one centimetre of the distracted site. It is directly proportional to the age of the patient.\textsuperscript{15} The older the patient, the higher is the risk for delayed healing, need for revision bone grafting and nonunion. Therefore, if possible, surgery should not be delayed to adulthood.

The earliest lengthening of the metacarpal was carried out by Tajima in 1976 using an iliac crest graft in a single stage.\textsuperscript{18} Lengthening, either acutely or by callotasis, remains the mainstay of treatment in brachymetacarpia. The amount of lengthening can be estimated by the formula given by Aydinlioglu:\textsuperscript{1} first metacarpal = 0.67 × second metacarpal = 0.71 × third metacarpal = 0.78 × fourth metacarpal = 0.84 × fifth metacarpal. A dorsal approach is commonly used for the osteotomy.\textsuperscript{16}

Acute lengthening involves a transverse osteotomy at a mid-metacarpal position and distraction intraoperatively,
maintained by the use of autologous iliac crest bone graft and Kirschner wires or mini-plates.\textsuperscript{14} This procedure is limited by soft tissue tension and up to 7–10 mm of length achieved. Callotasis can be carried out either in a single stage or two stages.\textsuperscript{14} In single-stage callotasis (Fig. 5), a transverse osteotomy of the metacarpal shaft is followed by gradual lengthening by the use of a unilateral external fixator, usually at the rate of 0.5 to maximum 1 mm/day in the beginning which is usually started between day 4 and 7. The rate of distraction is evaluated every week with radiographs and the rate altered according to the clinical tolerance as well as the quality of callus formation. In two-stage callotasis, after the initial osteotomy, distraction is carried out at a faster rate of 2 mm/day till the desired length has been achieved. At this stage, the second surgery entails autologous bone grafting, commonly from the iliac crest, at the site of lengthening followed by removal of the external fixator and replacement by internal fixation either by Kirschner wire or mini-plate. This procedure is associated with a greater rate of complications owing to the fast distraction due to joint dislocations, neurovascular disturbance and cutaneous problems.

The decision when two different metacarpals are involved in the same hand can be tricky. When two different metacarpals are involved, the long ray is preferentially elongated because the knuckle line of the third ray attributes more cosmesis to it as compared to that of the fourth or fifth ray.\textsuperscript{18}

\textbf{Brachymetatarsia}

Surgery is usually performed after the age of 12 years in females and 14 years in males because the growth of the metatarsals is near completion and the postoperative rehabilitation and care can be administered more efficiently in an adolescent.\textsuperscript{42} As mentioned above, children have a lower healing index with reduced rates of complications compared to adults.\textsuperscript{32}

Acute or gradual lengthening remains the mainstay of treatment for brachymetatarsia. McGlamry and Cooper reported the first metatarsal lengthening in 1969,\textsuperscript{46} Single-stage lengthening can be carried out with the use of autologous bone grafts from sites such as the iliac crest, calcaneum, fibula, tibia or adjacent metatarsal.\textsuperscript{47–49} A dorsal approach is also commonly used for the osteotomy. Kirschner wire or plate is commonly used to stabilize the graft. Synthetic spacers have been used in place of bone grafts in single-stage lengthening.\textsuperscript{50,51} The spacer can be made into a chevron shape to increase

\begin{figure}[h]
\centering
\includegraphics[width=\textwidth]{figure4.png}
\caption{An adolescent girl with severe shortening of the fourth metatarsal bone (as in Fig. 2) is shown before (A, C) and after gradual bone lengthening (B, D). Note the malposition of the metatarsal head in the lateral projection (arrow, C) and restoration of length after surgery (arrow, D).}
\end{figure}

\begin{figure}[h]
\centering
\includegraphics[width=\textwidth]{figure5.png}
\caption{A lengthening procedure with an external fixator in brachymetacarpia is shown. Note the shortening of the ring finger in relation to the long finger.}
\end{figure}
stability and prevent displacement. Increased lengthening can be achieved in these cases by adjunct soft tissue procedures for tendon and skin to accommodate the increased length.

Single or two-stage gradual lengthening can be achieved by callotasis. This is advocated for cases which require lengthening for more than 12–15 mm. Kirschner wires can be used to stabilize the MTP joints during distraction. In single-stage lengthening, distraction is started at 0.5 to 1 mm/day and then adjusted according to the weekly radiographs and clinical tolerance of the patients as mentioned before. In two-stage lengthening, the distraction is carried out at a faster rate 2 mm/day and after the desired length is achieved, bone grafting is carried out at the lengthening site and external fixation is replaced by internal fixation using wire or plate. Ilizarov semi-circular ring or, more commonly, unilateral fixators can be used for distraction.

Sliding lengthening osteotomies without the use of bone graft have been reported but are limited by the amount of lengthening that could be achieved. Shortening of the adjacent metatarsal or proximal phalanx has also been suggested to reduce the lengthening of the affected metatarsal. Syndactylization can also be offered as a treatment if cosmesis is not of concern to the patient. The ray is disarticulated from the metatarsal which can undergo syndactylization to the adjacent metatarsal. This procedure renders the affected ray non-functional.

Outcomes of surgery

Brachymetacarpia

Six studies reported the outcome following lengthening surgeries for brachymetacarpia. All the studies reported improvement in appearance and hand function as well as range of motion (ROM) of the MCP joint. The rate of lengthening varied from 0.1 mm/day to 0.7 mm/day. Total duration of distraction ranged between 32 and 49 days. The mean lengthening was 18.75 mm (10–28 mm). The healing index and consolidation time varied between 49.6 to 83.1 days/cm and 38.0 to 82.1 days respectively. A direct relationship between healing index and age of the patient was noted, with healing index increasing with increasing age. Body Image Quality of Life Index (BIQLI) and Limb Deformity modified Scoliosis Research Society (LD-SRS) scores were used to quantify the outcome in one of the studies which showed an improvement over the preoperative scores. Although, apart from these quantitative scores, all studies showed a subjective satisfaction on the part of the patients in the postoperative period. The rate of complication noted was 36.6% (0–100% range).

Brachymetatarsia

Twenty-two studies reported the outcome following lengthening surgeries for brachymetatarsia. All the studies reported improvement in cosmesis and reduction of pain while walking. ROM of the MTP joint was reported to be reduced in two studies but this did not affect the quality of life of the patients. The rate of lengthening varied from 0.2 mm/day to 0.8 mm/day. Total duration of treatment ranged between 53 and 173 days. The mean lengthening was 15.9 mm (5.0–32.5 mm). The healing index and consolidation time varied between 36 to 127 days/cm and 26 to 111 days respectively. A direct relationship between healing index and age of the patient was noted, with healing index increasing with increasing age. American Orthopaedic Foot and Ankle scores (AOFAS) were used to quantify the outcome in three of the studies. The mean final AOFAS score was excellent in all the patients included (> 85 points). The rate of complication noted was 19.5% (range, 0.0–48.1%).

Complications of surgery

Brachymetacarpia

Acute lengthening is limited by the soft tissue tension and can lead to neurovascular complications if more than 10 mm lengthening is targeted. Nonunion, delayed union and loss of correction are other complications associated with it. Pin tract infection and stiffness are the common complications associated with callotasis. Delayed union, early consolidation, fracture, MCP joint dislocation and deformity of the regenerate are other complications associated with callotasis.

Brachymetatarsia

Masada et al indicated that complications occur when lengthening is carried out to beyond 40% of the original length of the metatarsal. Acute lengthening is mainly associated with complications related to neurovascular compromise owing to excessive lengthening. Other complications associated are prolonged immobilization, stiffness, loss of correction, delayed union and nonunion.

Complications in gradual lengthening may be due to the use of an external fixator or the increased length achieved in this process. Stiffness of the adjacent joints, pin tract infection and MTP subluxation are the common complications noted. Other complications noted are delayed union, early fusion, deformity or fracture of the regenerate.

Summary

Brachymetacarpia and brachymetatarsia are rare congenital presentations in the paediatric and adolescent age groups. In the presence of impairment of hand function,
surgery for the correction of the deformity is worthwhile. In brachymetatarsia, presence of pain and toe deformities leading to painful callousities and difficulty in using footwear should be absolute indications for intervention. Cosmosis remains a relative yet important indication for surgery in both these conditions, even more so in brachymetacarpia. Multiple lengthening options are available for each of the conditions associated with inherent complications. Management should be tailored to the patients’ individual deformity and expectations. Individualization of surgical techniques and patient selection criteria can result in a cosmetic, functional result that is satisfactory to the clinician and the patient.

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