Workshop report

221st ENMC International Workshop:

Foot Surgery in Charcot-Marie-Tooth disease

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Highlights

- Foot deformities are frequently observed in Charcot-Marie-Tooth disease patients.
- Surgical management is variable among centres.
- During the workshop current surgical approaches were reviewed.
- Recommendations for foot surgery in CMT patients were agreed by the participants.

1. Introduction

Nineteen clinicians with a specialist interest in Charcot Marie Tooth disease (CMT) (5 adult and 2 paediatric neurologists, 9 orthopaedic surgeons, 2 therapists, 1 specialist in rehabilitation medicine) and three patient representatives from 8 countries (UK, Italy, Netherlands, Belgium, Germany, Sweden, US and Australia) gathered for the 221st ENMC International workshop on foot surgery in Charcot-Marie-Tooth (CMT) disease on 10-12 June 2016 in Naarden, The Netherlands. Foot surgery is often performed in patients with CMT as foot deformities are frequent complications of this disease. Several procedures have been described to correct severe foot deformities in CMT and surgical approaches appear to vary between centres in the same country and between different countries. The aims of the workshop were to review the current surgical approaches, to agree on some basic principles and recommendations for foot surgery in CMT patients (adults and children) and to identify areas for further research.

2. Background to workshop

CMT is the most common inherited neuromuscular disorder, with an overall prevalence of 1 in 2500 individuals [1]. It is a genetically heterogeneous disease with over 90 causative genes
identified so far. Clinically it is usually characterised by slowly progressive distal weakness and sensory loss in the lower limbs progressing in a length dependent fashion to involve the upper limbs. Onset usually occurs in the first decade of life. Foot deformities, such as pes cavus and toe deformities, are frequently observed with a reported prevalence range from 60-90% [2-5].

Currently there are no available pharmacological treatments for CMT; however research is ongoing and various therapeutic approaches are under investigation. Symptomatic management is currently the mainstay of treatment and includes physiotherapy, exercise advice, provision of orthoses (in-shoe and ankle-foot orthoses), fatigue management and pain management. Non surgical approaches for foot deformities include triceps surae stretching exercises to maintain ankle dorsiflexion range of movement, joint mobilisation, muscle strengthening exercises, support of foot alignment with foot orthoses and use of ankle foot orthoses to reduce foot drop and correct ankle instability.

The aims of surgical intervention are usually to re-align the joints, to correct the bony deformities and imbalance of the muscles [6]. Surgical decisions are usually determined by the age of the patient, the severity of the deformity, the need to prevent further deterioration, and when pain and decreased function are not corrected by the non surgical measures mentioned above. Several procedures have been described for the correction of foot deformities. They include soft tissue procedures, such as plantar fascia release, tendon transfers and tendon lengthening, corrective osteotomies and fusion procedures, such as triple arthrodesis. No single surgical procedure can achieve all these goals simultaneously, and surgical decisions are usually individualised [6]. The long-term results of surgical procedures in CMT patients have been addressed in just a few studies [6-8], so evidence regarding optimal surgical management is lacking. It is therefore difficult to guide decisions on what surgery an individual patient should have and when they should have the surgery. Guidelines are urgently needed and hence that rationale for this workshop.
3. Workshop - Pre workshop pilot work

3.1 Foot deformity and surgery frequency in CMT and related disorders

During the workshop Mary Reilly (London, UK) presented data on foot deformities (pes cavus, toe deformities and pes planus) and foot surgery (tendon transfer, Achilles tendon lengthening, ankle joint fusion, osteotomy and toe straightening) collected as part of a minimal dataset included in an ongoing large Natural History Study on Charcot Marie Tooth and related disorders. This natural history study is being conducted by the Inherited Neuropathy Consortium (INC) which is an NIH funded consortium including 19 sites (16 in the USA, and one each in the UK, Italy and Australia) specialised in translational research in CMT (INC Principal Investigator (PI) is Michael Shy, University of Iowa) [9].

Data obtained from 2706 patients recruited between August 2009 and December 2014 showed that foot deformities were reported in 1914 (71%) patients with a diagnosis with CMT and related disorders (including Hereditary Neuropathies with liability to pressure palsies (HNPP), Hereditary Motor Neuropathy (HMN) and Hereditary Sensory Neuropathy (HSN)), and were more common in patients with a diagnosis of CMT (1443/1953 (74%)) compared to other subtypes (HNPP, HMN and HSN). Pes cavus and toe deformities were more commonly reported in CMT patients, 1089/1953 (56%) and 534/1953 (27%) respectively. Pes planus was only reported in a minority of CMT cases 150/1953 (8%). From the database it was ascertained that 30% of the patients with foot deformities (574/1914) had undergone corrective foot surgery. A variety of procedures which included bone and soft tissues procedures were performed [10].

3.2 Surgical practice survey

Matilde Laurá (London, UK) presented the results of a pilot study performed by the INC in preparation for the workshop. Discussions among INC workshop neurologists with large
CMT practices (including Reilly, Pareyson, Shy, Laurá) prior to this workshop suggested that there are differences in the types and timing of surgeries offered between different centres and between different countries. The aim of the pilot study was to determine whether current surgical approaches to management of orthopaedic complications in CMT are variable among specialised centres. The study included a survey designed by a foot and ankle surgeon (Dishan Singh, London, UK) addressed to experienced CMT orthopaedic surgeons performing surgical procedures for foot deformities in CMT patients attending centres participating in the INC. In brief, surgeons were asked what procedures they would commonly carry out in two clinical scenarios (one adult and one paediatric). Sixteen orthopaedic surgeons (from UK, Italy, Australia and US) were surveyed and the results showed a great variability of surgical approach for correction of pes cavus. Overall in both scenarios the combination of procedures selected by the surgeons varied considerably, with only two surgeons selecting a similar combination of procedures to correct pes cavus for a paediatric case (calcaneal osteotomy, peroneal tendon transfer, first metatarsal osteotomy and plantar fascia release) and none of the surgeons selecting an identical combination of procedures for an adult case. The pilot survey confirmed the perception of the variability of surgical approaches to management of orthopaedic complications in CMT even among centres specialised in CMT and highlighted the need for further research [10].

4.0 Workshop – Indications, pre surgical assessment and timing of surgery

4.1 Background, literature review and personal experience

Currently there are no specific guidelines for foot surgery for children and adults. Viola Altmann (Nijmegen, the Netherlands) presented data on an extensive literature review up to January 2015 on surgical guidelines for foot surgery in CMT. Currently there are no guidelines, Cochrane review or randomised controlled trials on timing of surgery or on type of patients to be referred for surgery. Current evidence is based on case series with small numbers of CMT patients per study. The review concluded that there is limited evidence on
timing of surgery but that surgical procedures are best performed at an early stage, before severe deformities occur, although the risk of recurrence of deformities requiring further surgery is possible. Furthermore, based on limited evidence (level C), patients can reasonably expect a stable, straight foot, no need for walking aids or orthopaedic shoes and no pain after an operation. There is also limited evidence that gait pattern and walking distance can improve. There is no evidence on the effect of growth on the timing of surgery or the preliminary assessment that should be done before surgery.

Paul Gibbons (Sydney, Australia) reviewed the literature specifically on timing of surgery in children with CMT and currently there is no consensus. He presented data on personal experience over 10 years (2006-2016) from 22 CMT paediatric surgical cases. Indications for surgery were severity and progression of deformities, intolerance of orthosis, pain, ankle instability and falls.

Josh Burns (Sydney, Australia) outlined current indications for surgery in children which include painful and severe foot deformities and the prevention of fixed deformities, recurrent ankle sprains, foot drop, poor balance and intolerance of orthoses. He also presented current assessment protocols for foot deformity requiring surgery including the Foot Posture Index, observational and instrumented gait analysis including pedobarography before and after surgery.

Gita Ramdharry (London, UK) presented a clinical reasoning model for timing of surgery in adults including complexity of presentation and failure of conservative measures, for example inability to correct or support the deformity with orthosis, poor tolerance, risk of musculoskeletal injury and limited potential to increase range of movement.

Dishan Singh discussed assessment of foot deformities in adults and the use of Coleman block test to evaluate how correctible a deformity is. This is not a validated test but there was agreement among the surgeons that the test is useful to evaluate how correctible is the hindfoot but it used as is not an outcome measure. He also highlighted the importance of the
evaluation and assessment of other joints, e.g. knees and hips, before performing foot and ankle surgery. He identified some red flags where surgery needs to be considered including recurrent stress fractures of the fifth metatarsal bone.

4.2 Workshop agreements - Indications, pre surgical assessment and timing of surgery

4.2.1 Indications

It was agreed that surgery is usually indicated when conservative measures for foot deformity such as orthosis and physical therapy interventions have failed. Conservative treatment failure is considered when deformities cannot be supported or corrected by orthosis and when pain becomes intolerable.

4.2.2 Pre surgical Assessment

It was agreed that if surgery is being considered for children or adults, the patient should be assessed in a multidisciplinary setting. The multidisciplinary team may vary between centres but should ideally include an orthopaedic surgeon, an adult or paediatric neurologist, and a therapist. A variety of therapists may be involved in managing patients with CMT including physiotherapists, podiatrists and orthotists and in some centres rehabilitation physicians and occupational therapists are actively involved. It was further agreed that surgery should ideally be undertaken by a foot and ankle orthopaedic surgeon with expertise on CMT.

4.2.3 Proforma for referral for surgery

The workshop agreed that a proforma with essential background information be provided by neurologists at the time of referral of a patient with CMT to an orthopaedic surgeon to accompany the referral detailing the problem. This should include details of patients’ diagnosis (including genetic if known), severity (e.g. CMT Neuropathy Score), prognosis, specific clinical features that could influence surgery (vocal cord palsy, sleep apnoea, diaphragmatic weakness or other respiratory difficulties, severe sensory loss, healing
difficulties, recurrence of stress fractures, hip displacement). A proforma (Proforma 1a, Appendix 1) was agreed during the workshop and is designed to accompany the standard referral letter which will vary between centres. A shortened version of the proforma was also agreed for straightforward patients (Proforma 1b, Appendix A).

4.2.4 Timing of surgery

There is no clear evidence on optimum timing of surgery in children and the risk of deformity recurrence as well as the progressive course of the condition need to be taken into consideration in planning surgery. It was agreed that further prospective studies are needed before guidelines on timing of surgery in children can be developed.

For adult cases it was agreed that patients should be referred for a surgical opinion when conservative measures have failed, when the deformity has become rigid and severe and when there is risk of developing musculoskeletal injury as result of the foot deformities.

5. Workshop - What surgery

5.1 Background, literature review and personal experience

Current surgical procedures

The various current surgical procedures performed in CMT patients were discussed in detail by the orthopaedic surgeons present at the workshop. The surgical procedures discussed were: soft tissue procedures (Achilles tendon lengthening, gastrocnemius lengthening, plantar fascia release, transfer of peroneus longus tendon to peroneus brevis tendon, tibialis posterior tendon transfer, Robert Jones procedure, lesser toe tendon transfer); osteotomies (calcaneal osteotomy, midfoot osteotomy, first metatarsal osteotomy); fusion (ankle arthrodesis, triple arthrodesis, isolated hindfoot arthrodesis (e.g. Lambrinudi), midfoot arthrodesis and lesser toe fusion) [11]. Each surgeon outlined the role of each technique in Charcot Marie Tooth disease, presenting data from both literature reviews and data/cases from personal experience.
Some techniques not routinely performed in CMT patients such as peroneus longus tendon lengthening, tibialis posterior lengthening and tibialis anterior tendon transfer, ankle arthroplasty and the use of external fixators were also reviewed and discussed.

5.2 Workshop agreements – What surgery

5.2.1 Aim of surgery

It was agreed that the main aim of surgery is to achieve a stable, well balanced foot and to improve pain. The decision on timing and type of surgery should be taken in a multidisciplinary team, as described above.

5.2.2 Acceptable surgical options

During the workshop, despite the lack of evidence for specific surgical guidelines for foot deformity in CMT, it was agreed after review of the literature and review of contemporary practices by different participants that some procedures were deemed to be acceptable in the correction of foot deformities. The importance of correcting muscle imbalance in foot surgery for CMT was highlighted.

It was agreed that:

a. tibialis posterior transfer should be performed when severe foot drop is present and the varus hindfoot deformity is still flexible. Tibialis posterior tendon transfer can be combined with stabilising/corrective bony procedures (such as osteotomies and fusions) in order to enhance ankle dorsiflexion and improve muscle imbalance.

b. Robert Jones procedure (transfer of the Extensor Hallucis Longus to the neck of the first metatarsal) in isolation is acceptable in paediatric cases when the deformity is flexible but in adult cases the consensus was for a combination of the procedure with metatarsal osteotomy.

c. calcaneal osteotomy is effective in correcting hindfoot and is usually performed in combination with other techniques.
d. dorsiflexion osteotomy at the base of the first metatarsal is recommended for correction of an isolated fixed plantarflexed first ray, usually after hindfoot correction.

e. lesser toe fusions are usually performed in combination with other procedures and when conservative measures have failed.

f. there was variation in opinion on the timing of triple arthrodesis with some surgeons performing the procedure earlier than others. However all the surgeons agreed that ankle fusion is needed when disabling ankle arthritic changes are present.

5.2.3 Suggested procedures that should be avoided

Some procedures were deemed not to be suitable for CMT patients with muscle imbalance and foot deformity

a. Isolated peroneus longus tendon lengthening is not recommended.

b. Tibialis posterior tendon lengthening is not recommended.

c. Tibialis anterior transfer should not be routinely considered.

c. Isolated plantar fascia release is not recommended.

d. External fixators are not advisable in the correction of foot deformities as muscle balance also has to be restored.

e. Ankle arthroplasty is not recommended.

f. In children arthrodesis is not recommended.

5.2.4 Record of surgical procedures performed

The workshop also agreed that a proforma listing the individual surgical procedures performed during surgery should be completed by the surgeon for the treating neurologist for CMT. The workshop agreed the list of procedures to be included in the proforma (Proforma 2, Appendix B).

5.2.5 Questions needing further research
It was agreed that a comparison of surgical techniques is needed with both short term and long
term follow up. Furthermore, it was agreed that developing a standardised follow up protocol
to be implemented internationally would be an achievable initial method to allow proper
comparison of surgical techniques and to allow long term follow up of outcomes. It was also
agreed that retrospective studies on patients previously operated to evaluate outcome
depending on type of surgical procedure would be valuable. Ideally both prospective studies
and open trials addressing specific questions in children and adults are needed. It was clear
from discussions that traditional randomised trials will not be possible as surgeons develop
expertise in a particular technique or combinations of techniques and it would be unreasonable
and not desirable to ask surgeons to randomise the procedures they perform but outcomes can
be compared between sites that use different techniques. Long term follow up of outcomes in
a standardised fashion will be the best way to see which procedures or combination of
procedures work best.

6. Workshop - Post operative care

6.1 Background and discussion

In the workshop there was a wide ranging discussion regarding post operative issues including
the optimum length of immobilisation post operatively, which outcome measures should be
used, what type of rehabilitation should happen in the immediate and the long term post
operatively and how long and what type of follow up is needed. Discussion also took place on
the need for more accurate prognosis for surgical procedures.

6.2 Workshop agreements – post operative care.

Broad guidance for a follow up protocol was agreed among participants. Ideally the protocol
should include: impairment scales, quality of life questionnaires, patient reported outcome
measures (PROM), pain questionnaires, Foot Posture Index, alignment and angles of the foot,
X-ray, gait analysis such as pedobarography, muscle MRI, orthopaedic scales (e.g. Foot
Function Index, Manchester-Oxford questionnaire, Maryland foot score) and scales to assess balance. Further work is needed to refine and implement the follow up protocol.

7. Conclusions

The workshop was agreed by all participants to have been very successful. All the aims of the workshop were met, i.e. to review the current surgical approaches for correction of foot deformities in CMT, to agree some basic principles and recommendations for foot surgery and follow up in CMT patients. During the workshop a number of key principles were agreed:

1. The decision on timing and type of surgery should be taken in a multidisciplinary team;
2. The main aim of surgery is to achieve a stable well-balanced and pain free foot;
3. A standardised follow up protocol needs to be developed to adopt in the clinical setting and which will be suitable for future research purposes and to allow proper comparison of surgical techniques and proper long term outcomes.

A working group was formed to take the actions identified forward.

Workshop participants

Per Henrik Agren (Stockholm, Sweden)
Viola Altmann (Nijmegen, the Netherlands)
Jonathan Baets (Antwerp, Belgium)
Peter Briggs (Newcastle upon Tyne, UK)
Joshua Burns (Sydney, Australia)
Karen Butcher (Christchurch, UK)
Luca Gaiani (Imola, Italy)
Filippo Genovese (Modena, Italy)
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