CASE REPORT

Essential timing of orthopaedic treatment in children with Ehlers-Danlos syndrome arthrochalasia type: a case report

[version 1; peer review: 1 approved, 1 approved with reservations]

Maarten J. van Rooij¹, Heleen Staal², Merel Klaassens³, Lodewijk Van Rhijn², Connie T.R.M. Stumpel⁴

¹Department of Orthopaedic Surgery, Maxima Medisch Centrum, Eindhoven, 5504 DB, The Netherlands

²Department of Orthopaedic Surgery, Maastricht UMC+, Maastricht, 6229 HX, The Netherlands

³Department of Pediatrics, Maastricht UMC+, Maastricht, 6229 HX, The Netherlands

⁴Department of Clinical Genetics and School for Oncology & Developmental Biology (GROW), Maastricht UMC+, Maastricht, 6229 HX, The Netherlands

V1 First published: 06 Feb 2014, 3:39
<https://doi.org/10.12688/f1000research.3-39.v1>
 Latest published: 06 Feb 2014, 3:39
<https://doi.org/10.12688/f1000research.3-39.v1>

Abstract

Objective: This case report presents the orthopaedic impact of arthrochalasia-type Ehlers-Danlos syndrome (EDS) (former type VIIA and B) in a young girl.

Methods: The characteristics of EDSs are skin abnormalities and tissue fragility with orthopaedic consequences including joint hypermobility and dislocations. In EDS arthrochalasia type (former type VIIA and B) severe general hypotonia and congenital bilateral hip dislocation are distinctive symptoms. This type of EDS is less common than other types and just a few cases have been reported.

Results: This report describes a case of a 3-year old girl with EDS arthrochalasia type and bilateral high hip dislocation, spontaneous shoulder dislocation and scoliosis as orthopaedic consequences.

Conclusions: Orthopaedic consequences in these cases can be severe so conservative or surgical treatment will be necessary to reduce disability in later life. Timing of either conservative or surgical intervention is essential. According to the literature, most conservative treatments fail. Also, surgical treatment is rarely effective in most of the cases. Both conservative and surgical treatment should be considered carefully in the treatment of EDS arthrochalasia type, each of which has pros and cons. The best treatment depends on the prognosis and current situation of the child.

Keywords

Ehlers-Danlos syndrome, arthrochalasia, orthopaedic surgery, treatment

Open Peer Review

Approval Status

	1	2
version 1
06 Feb 2014	view	view

1. **Patrick Calders**, Ghent University, Ghent, Belgium

2. **Zbigniew Gugala** , University of Texas Medical Branch at Galveston, Galveston, TX, USA

Any reports and responses or comments on the article can be found at the end of the article.

Corresponding author: Maarten J. van Rooij (mj.vanrooij@mmc.nl)

Competing interests: No competing interests were disclosed.

Grant information: The author(s) declared that no grants were involved in supporting this work.

Copyright: © 2014 van Rooij MJ *et al.* This is an open access article distributed under the terms of the [Creative Commons Attribution License](#), which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited. Data associated with the article are available under the terms of the [Creative Commons Zero "No rights reserved" data waiver](#) (CC0 1.0 Public domain dedication).

How to cite this article: van Rooij MJ, Staal H, Klaassens M *et al.* **Essential timing of orthopaedic treatment in children with Ehlers-Danlos syndrome arthrochalasia type: a case report [version 1; peer review: 1 approved, 1 approved with reservations]**

F1000Research 2014, 3:39 <https://doi.org/10.12688/f1000research.3-39.v1>

First published: 06 Feb 2014, 3:39 <https://doi.org/10.12688/f1000research.3-39.v1>

Introduction

Ehlers-Danlos syndrome (EDS) is a well known inherited connective-tissue disorder characterized by general joint hypermobility, tissue fragility and skin abnormalities. Six subtypes are described which show clinical overlap with each other and other syndromal connective tissue disorders (e.g. Larsen syndrome)^{1,2}. EDS arthrochalasia type (former EDS type VII-A and VII-B) can be distinguished from other types by the presence of mild dysmorphic features (in particular hypertelorism and micrognathia), congenital bilateral hip dislocation, multiple other recurrent (sub)dislocations and severe muscular hypotonia. This type of EDS is very rare with only 27 cases described to-date¹.

In this case report the history of a three year old girl with EDS arthrochalasia type is described. This girl has been described previously¹.

Medical history

A 6 year old Caucasian girl, known in the paediatric, clinical genetic and orthopaedic outpatient clinic since birth, was treated for severe skin problems, hyperlaxity of her joints and hypotonia. These congenital symptoms were initially thought to be a part of Larsen syndrome, because of the facial features and hypotonia. However, this would not explain the severe skin problems. At the first visit to our outpatient clinic, when the girl was four months, a bilateral hip dislocation in 90° abduction was seen. Manual closed reduction was considered a possibility. The shoulders dislocated spontaneously with maximum anteversion. X-rays showed high dislocations of the both hip joints and a thoracolumbar scoliosis (Figure 1 and Figure 2). At the age of six months EDS arthrochalasia type was suspected and confirmed by Sanger sequencing, which showed a *COL1A2* mutation.

Initially, the hip dislocation was not treated with a harness because of the easy bruising and multiple lacerations of the skin, and the unknown origin of the syndrome. After confirmation of the diagnosis, surgical hip reduction in the near future was believed not to be the best intervention. The reason for this was the high chance that obtaining a stable reposition would fail, because of the severe connective tissue abnormalities. Given the severe hypotonia and multiple other dislocations, the prognosis for walking was small. In addition, the patient would have to wear a plaster cast for at least 12 weeks postoperatively, which would cause major skin problems

Figure 1. Radiographs of the girl taken at two years of age showing bilateral hip dislocation at an anteroposterior view (left) and Lauenstein view (right).

Figure 2. Radiographs of the girl taken at two years of age showing thoracolumbar scoliosis at an anteroposterior view (left) and lateral view (right).

and would probably further impair her motor development. For the same reason conservative treatment in terms of only a plaster cast was deemed unachievable. This decision was supported by findings in the literature showing that conservative treatment in these cases is not successful.

Follow-up of the patient showed a girl with a progressive equinovarus deformity of both feet (Figure 3–Figure 5) starting at the age of 1 year and 6 months. Due to her foot deformity, bilateral hip dislocation and hypotonia it was not possible for the girl to mobilize. Initially, the foot deformity was mild and easy to redress. After one year (at age 2 years and 6 months) it was no longer possible to redress the foot deformity, due to contractures and stiffness of the joints. A decision for surgical treatment was made in order to allow shoe wearing. A bilateral tenotomy of the Achilles tendon and a bilateral anticus transfer was performed. Extension of the flexor hallucis tendon of the right foot and a tenotomy of the flexor hallucis tendon of the left foot were also part of the treatment. Postoperative treatment consisted of a well-padded plaster cast for 6 weeks and appropriate footwear afterwards. After this treatment, a slight foot deformity remained (Figure 6), although the intention of treatment, which was to redress the deformity in her feet so that they would fit shoes, was achieved.

Figure 3. Radiographs of the girl taken at two years of age showing a preoperative equinovarus deformity and several joint dislocations of the left foot at an anteroposterior view (left), lateral view (middle) and a 3/4 view (right).

Figure 4. Radiographs of the girl taken at two years of age showing a preoperative equinovarus deformity and several joint dislocations of the right foot at an anteroposterior view (left), lateral view (middle) and a 3/4 view (right).

Figure 5. Preoperative equinovarus deformity of the left foot and postoperative deformity reduction of the right foot of the girl at the age of two years and six months.

Figure 6. Postoperative deformity reduction of both feet of the girl at the age two years and eight months.

Discussion and consideration

In 1892, Tschernogobow was the first to describe joint dislocations due to ligamentous and capsular laxity⁴. Ehlers-Danlos syndrome is an inherited connective-tissue disorder with an autosomal dominant mode of inheritance³. General clinical symptoms are hypermobility of the joints, tissue laxity and skin abnormalities. There are six different subtypes, based on their clinical and genetic features. EDS arthrochalasia type is caused by mutations in the *COL1A1* (OMIM 130060) or *COL1A2* (OMIM 130060) gene, which causes

production of low quality type 1 collagen fibers. This EDS type distinguishes itself from the other types of EDS by the severity of the congenital bilateral hip dislocations, recurrent subluxations, subtle dysmorphic features, a severe muscular hypotonia, a soft velvety skin that is not hyperextensible and a 'criss-cross' patterning of the palms and soles^{1,3}.

Of all forms of EDS, approximately 90% are the classic and hypermobile subtypes (EDS I & III) and 5–10% is the vascular subtype (IDS IV). All the other subtypes are extremely rare. Only 27 cases of EDS arthrochalasia type are described in the literature^{1,2}.

A postnatal early diagnosis of EDS arthrochalasia type is important, because an early diagnosis will benefit follow-up and treatment. Important issues compromising development, such as the severe hypermobility with dislocations and hypotonia, can be addressed appropriately. Establishing a diagnosis can be difficult in the neonatal period, because of phenotypic overlap with other skeletal dysplasias, such as Larsen syndrome^{1,3}.

There is no consensus in literature on treatment of EDS arthrochalasia type. The little knowledge there is on treatment of the upper limbs tells us that neither orthotics nor surgical treatment are effective⁵.

With regard to the lower limbs, a choice between conservative treatment vs. surgical treatment is an important step in the treatment process. Conservative treatment, such as plaster casts, Pavlic bandages or orthotics, would be preferable, but they have a high chance of causing decubitus lesions, due to high pressures on a highly vulnerable skin. Therefore, these treatments are not favoured in patients with motor retardation. Also for lower limbs it seems, according to the literature, that conservative treatment is not effective. Surgical treatment is effective in only some cases in the treatment of congenital hip dislocations (Table 1)⁵. Only an iliac osteotomy with or without a varus and derotation femoral osteotomy shows stability in other hyperlaxity syndromes^{1,5}.

For the treatment of equinovarus deformities of the feet, which are common in EDS arthrochalasia type patients, treatment options are also described to be rarely effective⁶.

Although surgical procedures are invasive, they seem to be the only proven treatment to be effective. In this case a bilateral hip dislocation and an equinovarus deformity of both feet was seen and

Table 1. Results after treatment of a congenital hip dislocation (from Guinta et al.)⁵.

Treatment	No of cases	Result
Conservative treatment - orthosis/casts - closed reduction	16	No stable repositions achieved
Open reduction* - without osteotomy	10	A stable reposition was achieved in 1 out of 10 cases
Open reduction* - with osteotomy	6	A stable reposition was achieved in 2 out of 6 patients

*Several conservative treatment options preceded surgery

conservative treatment was considered not to be effective or to possibly be even harmful in this child. Therefore, a capsular and tendon release of the feet was carried out. Another treatment option for the feet could have been a subtalar release or a triple arthrodesis^{6,7}.

It is advisable to wait till the end of childhood for correction of the scoliosis deformity. In general, muscles will have gained strength during childhood, partly because of improvement in muscle tone. This most likely will result in an improvement of the scoliosis⁶.

Conclusion

EDS arthrochalasia type is rare. In all patients the dislocation of the joints combined with hypotonia is a serious problem, leading to severe disability. All interventions, both conservative and surgical, should be considered carefully. As with any treatment method, complications such as wound healing problems and decubitus, resulting from the poor quality of the collagen fibers, are a risk. The best treatment depends on prognosis and current situation of the child.

Consent

Written informed consent for publication of her clinical images was obtained from the parent of the patient.

Author contributions

MR, HS and MK conceived the study and prepared the draft of the manuscript. LR and CS revised the article critically.

Competing interests

No competing interests were disclosed.

Grant information

The author(s) declared that no grants were involved in supporting this work.

Acknowledgements

We thank the family of the patient for their cooperation.

References

1. Klaassens M, Reinstein E, Hilhorst-Hofstee Y, *et al.*: **Ehlers-Danlos arthrochalasia type (VIIA-B) – expanding the phenotype: from prenatal life through adulthood.** *Clin Genet.* 2011; **82**(2): 121–30.
[PubMed Abstract](#) | [Publisher Full Text](#)
2. Yen JL, Lin SP, Chen MR, *et al.*: **Clinical features of Ehlers-Danlos syndrome.** *J Formos Med Assoc.* 2006; **105**(6): 475–480.
[PubMed Abstract](#) | [Publisher Full Text](#)
3. Beighton P, De Paepe A, Steinmann B, *et al.*: **Ehlers-Danlos syndromes: revised nosology, Villefranche, 1997. Ehlers-Danlos National Foundation (USA) and Ehlers-Danlos Support Group (UK).** *Am J Med Genet.* 1998; **77**(1): 31–37.
[PubMed Abstract](#) | [Publisher Full Text](#)
4. Beighton P, Horan F: **Orthopaedic aspects of the Ehlers-Danlos syndrome.** *J Bone Joint Surg Br.* 1969; **51**(3): 444–453.
[PubMed Abstract](#)
5. Giunta C, Superti-Furga A, Spranger S, *et al.*: **Ehlers-Danlos syndrome type VII: clinical features and molecular defects.** *J Bone Joint Surg Am.* 1999; **81**(2): 225–238.
[PubMed Abstract](#)
6. Hamada S, Hiroshima K, Oshita S, *et al.*: **Ehlers-Danlos syndrome with soft-tissue contractures.** *J Bone Joint Surg Br.* 1992; **74**(6): 902–905.
[PubMed Abstract](#)
7. Trudgian J, Trotman S: **Ehlers-Danlos syndrome and wound healing: injury in a collagen disorder.** *Br J Nurs.* 2011; **120**(6): S10, S12, S14 Passim.
[PubMed Abstract](#)

Open Peer Review

Current Peer Review Status:

Version 1

Reviewer Report 16 April 2014

<https://doi.org/10.5256/f1000research.3512.r4502>

© 2014 Gugala Z. This is an open access peer review report distributed under the terms of the [Creative Commons Attribution License](#), which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited.

Zbigniew Gugala

Department of Orthopaedic Surgery and Rehabilitation, University of Texas Medical Branch at Galveston, Galveston, TX, USA

All available reports in the literature emphasize the extreme musculoskeletal challenges in the course and managements of patients with arthrochalasia-type (formerly type VIIA and VIIB; [OMIM 130060](#)) Ehlers-Danlos syndrome (EDS), and indicate no progress in effective therapy regardless of whether it is nonsurgical or surgical. All sorts of orthopaedic surgical interventions have been tried to improve patient gait and/or mobility, however, they all typically produce disappointing outcomes, thereby justifying surgery for only selected and specific indications in favor of physical therapy and/or bracing.

The case report by van Rooij *et al.* describes a patient with arthrochalasia-type EDS who has the characteristic severe musculoskeletal phenotype, specifically related to profound joint laxity and severe muscular hypotonia. The authors report a case of a girl with arthrochalasia EDS exhibiting phenotypic features typical for the disease (congenital bilateral hip dislocations, multiple and recurrent other joint sub- or dislocations, severe muscle hypotonia, velvety skin, facial signs) with the diagnosis confirmed genetically with a presence of *COL1A* mutation. Considering the protracted bilateral hip dislocation combined with severe muscle hypotonia any intervention, surgical or nonsurgical, had a very poor chance for tangible improvement of the patient's mobility. Furthermore, the skin fragility and abnormality posed significant risk for developing laceration, bruises, healing problems, thereby limiting all possible treatment options. Because the patient developed bilateral equinovarus foot deformity, and subsequent contractures and joint stiffness prevented a conservative treatment, the decision was made to surgically correct the foot deformity. Postsurgical cast treatment was uneventful, and ultimately a satisfactory outcome was achieved. This surgical intervention prompted the authors to discuss the challenges with any orthopaedic intervention for arthrochalasia-type EDS patients, and focus on the importance of identifying true surgical goals, while accepting high risk for complications.

The manuscript comprises a valuable reminder about the formidable treatment challenges that exist for these patients—the cases fortunately very rare.

There are, however, some inconsistencies and/or deficiencies in providing important and complete information about the case. The patient's age is contradictorily specified (i.e., 3 years old in the Introduction versus 6 years old in the Medical history section), and it remains unclear if any - and if so, what - treatment was performed to address the congenital bilateral hip dislocation, and how this was tolerated by the patient. The reader would also benefit from a more thorough discussion on the benefits and risks of early versus late surgical treatment, and the specific goals of surgery, if it is indeed indicated for these patients.

Competing Interests: No competing interests were disclosed.

I confirm that I have read this submission and believe that I have an appropriate level of expertise to confirm that it is of an acceptable scientific standard, however I have significant reservations, as outlined above.

Reviewer Report 03 April 2014

<https://doi.org/10.5256/f1000research.3512.r4333>

© 2014 Calders P. This is an open access peer review report distributed under the terms of the [Creative Commons Attribution License](#), which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited.

Patrick Calders

Department of Physical Therapy and Motor Rehabilitation, Ghent University, Ghent, Belgium

This is a well written case study which I 'Approve' without any revision necessary. It is clear and the fundamental information needed to understand each step is well described.

Competing Interests: No competing interests were disclosed.

I confirm that I have read this submission and believe that I have an appropriate level of expertise to confirm that it is of an acceptable scientific standard.

The benefits of publishing with F1000Research:

- Your article is published within days, with no editorial bias
- You can publish traditional articles, null/negative results, case reports, data notes and more
- The peer review process is transparent and collaborative
- Your article is indexed in PubMed after passing peer review
- Dedicated customer support at every stage

For pre-submission enquiries, contact research@f1000.com

F1000Research