update ERN CRANIO guideline Robin Sequence

Philippe Pakter <pakter@pierrerobineurope.com> Thu, Apr 22, 2021 at 10:48 PM
To: "I.M.J. Mathijssen" <i.mathijssen@erasmusmc.nl>
Cc: "R. de Ruiter" <r.deruiter.1@erasmusmc.nl>, "O.K.C. Spivack" <o.spivack@erasmusmc.nl>, Anne-Laure Aslanian <anne-laure.aslanian@eurordis.org>, Neil Russell <russell@pierrerobineurope.com>

Dear Irene,

I am writing in response to an email I received yesterday from ERN-Cranio Project Manager Renée de Ruiter informing me that ERN-Cranio has now begun developing Guidelines of Care for Pierre Robin Sequence.

In June 2018 I approached EURORDIS regarding ePAG representation in ERN-Cranio for the rare disease, Pierre Robin Sequence. Detailed discussions followed with various ERN-Cranio stakeholders including Mr. Matt Bolz-Johnson, Dr. Corstiaan Breugem, Mr. Gareth Davies and you, ERN-Cranio Coordinator Dr. Irene Mathijssen. At the end of this process you Irene, ERN-Cranio's Coordinator, formally and officially accepted Stichting Pierre Robin Europe, the organization I chair, into ERN-Cranio as an ePAG patient organisation.

In our application we expressed a specific interest in contributing to the development of Guidelines of Care for Pierre Robin Sequence. However in 2018, no work was apparently being carried out by ERN-Cranio to develop these Guidelines. Stichting Pierre Robin Europe therefore decided to take the initiative and contribute to the Pierre Robin Sequence community and to ERN-Cranio in a different way. Pierre Robin Sequence parents need clear, easy to understand, medically accurate, and sensitively written information about what PRS is, and what treatments are available. In practice it is very difficult to produce this - a document which is both medically accurate, and at the same time accessible to a non-healthcare audience. We noticed that the ERN-Cranio website offered parents a description of Pierre Robin Sequence of approximately 100 words. The description includes medical terms such as "hypoplasia" and "glossoptosis". The vast majority of Pierre Robin Sequence parents, we believe, will not understand what these words mean. Not only is the information limited in terms of quantity, but the information which is provided can do nothing more than leave a Pierre Robin Sequence parent confused:
https://ern-cranio.eu/diagnoses/cleft-lippalate-and-orodental-anomalies/robin-sequence/

Therefore Stichting Pierre Robin Europe, working together with ERN-Cranio affiliated doctors, nurses, surgeons and orthodontists, including Corstiaan Breugem, Peter A. Mossey, Sirpa Railavo, Pia Vuola, Ulla Elfving-Little, Elina Swan, Janne Suominen, as well as parents, Pierre Robin Sequence patient advocacy groups, and Pierre Robin Sequence Facebook groups, in the EU and beyond, produced an easy to read and medically validated information document for Pierre Robin Sequence parents. The document went through over 20 revisions; in its late stage drafts, ERN-Cranio's Dr. Corstiaan Breugem called it "great work". ERN-Cranio's Dr. Peter Mossey wrote that:

> "The combination of the original manuscript and the edits by Corstiaan have resulted in an excellent document. In the context of a parental leaflet on the topic, I find this to be a tremendously well written very comprehensive yet compact & useful information resource... I think it is that good".

There can be no question about the international standing or credentials of Dr. Peter Mossey - as a Professor, as the longstanding Director of the European Cleft Organisation, in terms of his international work with the WHO, not to mention his vast clinical experience - so this expert assessment from him carries great weight.

On the Stichting Pierre Robin Europe website this document has received thousands of visits, a striking fact when one considers that this is a rare disease. Pierre Robin Sequence parents throughout Europe and internationally have confirmed time and time again how helpful this document is. Many parents have told us how much they wish they had this document when their own Pierre Robin Sequence baby was born:
https://pierrerobineurope.com/parents/

Stichting Pierre Robin Europe's parent information document, produced in collaboration with ERN-Cranio healthcare providers, is a clear demonstration of the tremendous value which Pierre Robin Europe brings to ERN-Cranio as an ePAG patient organisation. And for this reason we sincerely looked forward to contributing to ERN-Cranio's Guidelines of Care for Pierre Robin Sequence in 2019.

However, once again, in 2019, no work was apparently carried out by ERN-Cranio to develop these critical Guidelines of Care for Pierre Robin Sequence.

As Stichting Pierre Robin Europe continued waiting to contribute to these Guidelines, we began pursuing another one of our organisation's formal patient advocacy goals, which is actually written into our organisation's deed of incorporation: to advocate for the development of a reliable Pierre Robin Sequence fetal ultrasonography screening

protocol, validated with prospective clinical trials, to reduce the incidence of airway emergencies, oxygen deprivation and death in the delivery room. To pursue this goal we contacted a respected surgeon and Pierre Robin Sequence expert from Harvard Medical School, Dr. Cory Resnick, who has been making progress in this particular area of Pierre Robin Sequence research. Dr. Resnick, who has been collaborating with ERN-Cranio's Dr. Maarten Koudstaal on a formal basis for years, granted Pierre Robin Europe an interview to explain to a non-medical audience the promising work they are doing at Harvard on the prenatal diagnosis of Pierre Robin Sequence. We successfully completed this project and published the interview on the Pierre Robin Europe website:
https://pierrerobineurope.com/prenatal-diagnosis-robin-sequence-dr-cory-resnick/

The response from Pierre Robin Sequence parents all around the world exceeded our wildest expectations. Pierre Robin Sequence parents in the EU and beyond are clearly hungry for high quality and accessible materials on this complex rare disease.

Stichting Pierre Robin Europe's Pierre Robin Sequence prenatal diagnosis interview is another clear demonstration of the unquestionable value which Stichting Pierre Robin Europe brings to the Pierre Robin Sequence patient community, and to ERN-Cranio. Once again this gave us reason to look forward to contributing to ERN-Cranio's Guidelines of Care for Pierre Robin Sequence in 2020.

However, once again, in 2020, no work was apparently carried out by ERN-Cranio to develop these very important Guidelines of Care for Pierre Robin Sequence. As Stichting Pierre Robin Europe's ePAG representative in ERN-Cranio, I received repeated assurances that the development of the Guidelines of Care for Pierre Robin Sequence would soon begin, and that we would soon be able to contribute to the development of these Guidelines. Unfortunately, we saw no movement.

We were officially admitted into ERN-Cranio in 2018. We are currently approaching the mid-point of 2021. I have finally received a message informing me that the long awaited development of these Guidelines of Care will begin. However neither I, nor Gareth Davies - ERN-Cranio formally promised the two of us that we would be contributing to the development of these Guidelines - neither one of us as official ePAG representatives has received any clear information on exactly what our involvement will be. We do not know exactly when this process will begin - or indeed if it already began. We have not been given an agenda for the development of these Guidelines. We have not been copied on any email correspondence concerning the upcoming schedule. We have not been told who will be involved. We have not been invited to a single meeting or video conference call. Indeed, we have not been permitted to have any type of meaningful or even symbolic involvement whatsoever - and it is an astonishing set of circumstances.

Stichting Pierre Robin Europe is an organization in ongoing contact with hundreds of Pierre Robin Sequence parents throughout Europe and all over the world. Stichting Pierre Robin Europe is an organisation with an official Medical Advisory Board which includes four medical doctors who are experts in Pierre Robin Sequence - and two of these four doctors also happen to be Pierre Robin Sequence mothers, an absolutely precious commodity, rare disease doctor-patients. In terms of our contributions, Stichting Pierre Robin Europe, as an ERN-Cranio ePAG patient organisation, has far exceeded the requirements of ERN ePAG representation - we have gone above and beyond the call of duty.

Recently the respected Elsevier medical journal, "Seminars in Fetal-Neonatal Medicine", decided to devote a full edition of their journal to Pierre Robin Sequence; all 15 articles in the edition will focus on this one rare disease. As Stichting Pierre Robin Europe's chairman, the editors invited me to contribute an article with the title: "A parent's view on the care of their baby with Robin Sequence". The article I have written offers visibility on the interests and concerns of Pierre Robin Sequence patients - which is necessary for providing patient centred care - which in turn contributes to better patient outcomes. One of the ERN-Cranio physicians who reviewed a draft of this article I wrote for "Seminars in Fetal-Neonatal Medicine" provided the following feedback:

> "Thanks, Phillipe - this is tremendous and it emphasises the need or should I say absolute imperative that we obtain and heed the patient / parent perspective in the approach to rare diseases. This certainly provides aspects that we do not read about in the textbooks."

An essential purpose of ePAG representation in the EU's ERN system is to ensure that the patients' voice is heard. The European Commission's Delegated Decision setting out criteria and conditions that European Reference Networks and healthcare providers must fulfill (2014/286/EU) is very clear about this, and makes it explicit: the ERNs are required to "empower and involve patients". Patient involvement is of paramount importance. So too is transparency - transparency in the way the ERNs, as recipients of EU funds, function and operate. The email I received from ERN-Cranio yesterday did not invite us to participate in the next meeting concerning these long awaited Guidelines of Care; instead the email indicates that you plan to contact patients/families via the ERN-Cranio centres to identify bottlenecks in care. Bottlenecks in care - is this the limit of patient involvement in the development of these Guidelines, which are so critical for us, the patients? We are limited to commenting on bottlenecks, without being permitted to contribute anything meaningful or participate in any way to a discussion of THE CARE ITSELF? Have you no interest in Pierre Robin Sequence parental concerns, treatment preferences, goals, fears, patient perspectives on burden of care, quality of life, nothing of this sort, just bottlenecks?

The information which ERN-Cranio provides on the ERN-Cranio website regarding Pierre Robin Sequence is woefully inadequate - approximately 100 words, including medical terms which we believe very few EU parents are likely to understand. Concerning the EU-funded Clinical Patient Management System, we at Stichting Pierre Robin Europe

know of not one single CPMS panel which has been opened up for a Pierre Robin Sequence baby since we were officially admitted to ERN-Cranio several years ago, in 2018. This leaves the Guidelines of Care. Stichting Pierre Robin Europe has been patiently waiting to meaningfully contribute to the development of these Pierre Robin Sequence Guidelines of Care ever since we joined ERN-Cranio years ago, back in 2018. Meaningful involvement is not the same as a vague opportunity to discuss "bottlenecks". This opaque and non-inclusive conduct on the part of ERN-Cranio cannot possibly be reconciled with the underlying principles and objectives of patient centred care, the ERN system, ePAG representation in the ERN system, or the 2011 Directive itself. I am confident that the European Commission, Members of European Parliament, EURORDIS, the European Patients' Forum, Vereniging Samenwerkende Ouder-en Patiëntenorganisaties, and countless other voices in the rare disease patient movement would agree with Stichting Pierre Robin Europe on this.

Before undertaking any further work on these Pierre Robin Sequence Guidelines of Care, which affect and concern patients, who should be treated as participants and stakeholders and members of a collaborative rare disease care team, I urge you to consider exactly what it is you are doing here, and what you are failing to do. In the process of developing Guidelines of Care we demand meaningful and active involvement right now; this should never have started without us, the patients.

We look forward to hearing from you at your earliest convenience.

Kind regards,

Philippe

--
Philippe Pakter
Stichting Pierre Robin Europe, voorzitter (chairman)
Member, EURORDIS, The European Organisation for Rare Diseases
Member, VSOP, Vereniging Samenwerkende Ouder-en Patiëntenorganisaties
PhD candidate, law: "Access to healthcare in Europe: the effectiveness of EU legislation in the context of rare disease patients"

On Wed, Apr 21, 2021 at 2:32 PM R. de Ruiter <r.deruiter.1@erasmusmc.nl> wrote:

> Dear Philippe,
>
> I would like to inform you that ERN CRANIO has started the development of a European Guideline on Robin sequence. The project is led by Erasmus MC as ERN CRANIO coordinating centre and involves all ERN CRANIO expert centres for RS. As part of this process we will contact patients/families via the ERN CRANIO centres involved to identify bottlenecks in organization of care for patients with Robin sequence.
>
> Best regards,
>
> Renée de Ruiter
>
> Project manager ERN CRANIO
>
> Erasmus MC Rotterdam