Epidermolysis Bullosa/European Reference Network
Registry meeting summary

Wed 8th June 2016, Dublin, Ireland

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Introduction

Dear friends and colleagues, we wish to extend a heartfelt thank you for the considerable contribution and positive energy that was contributed from all parties involved in the meeting. We hope that it acted as a stepping stone towards a collaborative approach to EB data collection and sharing that will positively disrupt research, care and patient involvement for many years to come. We believe that the shared experience and vision of this group can act, not just as a beacon of excellence to our own EB community, but to other rare and non-rare disease communities.

With best wishes,
Alan, Dmitri & Avril

Meeting Attendees

First name	Surname	Background	Country
Fiona	Aherne	DEBRA	Ireland
Leo	Bishop	DEBRA	Ireland
Fiona	Browne	Clinical	Ireland
Leena	Bruckner-Tuderman	Clinical	Germany
Pieter	Denorme	Clinical	Belgium
Veronika	Dvorakova	Clinical	Ireland
May	El-Hachem	Clinical	Italy
Jimmy	Fearon	DEBRA	Ireland
Cristina	Has	Clinical	Germany
Adrian	Heagarty	Clinical	UK
Victoria	Hedley	RD-Action	UK
Helmut	Hintner	Clinical/EB-CLINET	Austria
Alain	Hovnanian	Clinical	France
Alan	Irvine	Clinical/Irish Skin Foundation	Ireland
Greg	Johnston	eHealthIreland	Ireland
Avril	Kennan	DEBRA	Ireland
Eleanor	Krall	DEBRA	UK
Irene	Leigh	Clinical, BADGEM	UK
Mel	McIntyre	OpenApp	Ireland
David	McMahon	Irish Skin Foundation	Ireland
Ian	McNicoll	openEHR foundation/medical informatics	UK
Jemma	Mellerio	Clinical	UK
Cinzia	Pilo	DEBRA	Italy
Gabi	Pohla-Gubo	EB-CLINET	Austria
Ivan	Pristaš	PARENT	Croatia
George	Reynolds	OpenApp	Ireland
Holger	Storf	Medical informatics	Germany
Sara	Viegas	OpenApp	Ireland
Luciano	Vittozzi	EPIRARE	Italy
Dmitri	Wall	Clinical/Irish Skin Foundation/Medical Informatics	Ireland
Rosemarie	Watson	Clinical	Ireland
Giovanna	Zambruno	Clinical	Italy

Meeting welcome

Alan Irvine, on behalf of the Irish Skin Foundation registry development project and the proposed European Reference Network for Rare and Undiagnosed Skin Disease (ERN-Skin) eHealth group, welcomed the attendees.

Avril Kennan, on behalf of DEBRA, welcomed all and acknowledged that many in the room had already made great contributions to EB registry development. She described the need to work together towards a world-class EB registry and network, to support clinical care provision, research and advocacy.

Cinzia Pilo, on behalf of DEBRA International, also thanked those who had assembled and stressed the need for strong patient representation to drive success.

An informal, round-table introduction of attendees was then conducted. It was noted that the only solution for a successful international registry, is to develop a solution that embraces a network from the bottom-up. It was suggested that EB, with the excellent network facilitated by DEBRA, is the ideal condition to focus on, as a model of good practice for other rare skin diseases.

Presentations

Chaired by **Alan Irvine**, a number of excellent speakers gave their broad experience, to enable the EB community to develop high-quality patient registries, capable of communicating information, in the context of the emerging European Reference Networks (ERNs).

Luciano Vittozzi, on behalf of the EPIRARE project, delivered an excellent oversight of how this rare disease registry joint action could help direct development of an international EB registry. Of particular significance are the "common data elements" identified by the EPIRARE, with a view to defining core EB datasets. Luciano also gave an interesting overview of how EPIRARE principles had been applied in Italy's national Rare Disease Registry and highlighted EB data extracted from this registry.

Ian McNicoll, on behalf of openEHR, identified how openEHR is being utilised to build shared information models that technology can be built around. The strength of this approach, promoted by the PARENT (PAtient REgistries iNiTiative) Joint Action (JA), is the development of strong collaboration that results in interoperability across systems and national borders. It facilitates making consensus and dissent over information clear.

- This concept was likened to a means of enabling Universal Translation to ensure that data collected by registries is comparable.
- A further point was that openEHR, as a means to develop Electronic Health Records, is also a means to facilitate the collection of data from existing and developing clinical information systems.

Mel McIntyre, as the Director of OpenApp, the software company employed by the Irish Skin Foundation to develop its registries, discussed their work in developing a pan-European Cystic Fibrosis Registry. This review highlighted the practical difficulties of developing systems across international borders and meeting national security and privacy requirements. It also addressed the need to provide solutions with enough synergy to connect, while also meeting local requirements. He stressed the importance of the software company not owning the data but said that this should fall to service providers and/or patient organisations.

Ivan Pristaš, on behalf of PARENT JA, discussed the project's deliverables that could be utilised to facilitate the development of high-quality, cross-border patient registries. These include extensive registry

development guidelines and a registry of registries that can be used to identify registries and as an assessment and comparison tool for registries.

Dmitri Wall, presented a brief overview of the work of the eHealth group of the proposed ERN-Skin. This included reference to the work of **Holger Storf** and OSSE (Open-Source-Registersystem für Seltene Erkrankungen in der EU / Open Source Registry System for Rare Diseases in the EU) to create reusable software to develop rare-disease registries.

Victoria Hedley, on behalf of RD-Action, discussed the group's work to consolidate and formalise interactions between the RD and eHealth communities. This includes encouraging the development of IT platforms to meet the needs of RD networks, such as virtual care, research infrastructure development, genetic testing, data availability to patients, registry development and patient pathway definition. The planned role of the Connecting Europe Facility to provide an IT platform to support ERNs was discussed, as was a €1.2 million fund to support the creation of 3-4 patient registries for approved ERNs, on the basis of a competitive call.

Gabi Pohla-Gubo, on behalf of EB-CLINET, described their work to date in promoting the development of an international EB registry. This included the development of a proposed minimum dataset. The work undertaken with Peter van den Akker, Helmut Hintner and Marcel Jonkman to establish an EB Registry was also discussed.

Later in the day **Godfrey Fletcher**, on behalf of the Cystic Fibrosis (CF) Registry of Ireland gave his insights into the establishment of a national and European CF registry. This included an overview of the resources required to run it and the opportunities it has created. The major value of the CF Registry has been in the provision of rich longitudinal data that encourages and supports regional and international research. In addition it is recognised as a source of independently secured data that is invaluable in supporting advocacy, resource planning, quality control, health technology assessment (HTA) processes and in facilitating patient identification and recruitment for clinical trials.

Meeting outcomes

After breaking for lunch, there was discussion on how to progress the development of a connected network of EB registries. During this period there was considerable focus on the experience of national registries. Concerns, needs and potential benefits relating to an international network of registries were discussed. The major outcomes were as follows:

The need for a core dataset

- A core EB dataset should be established which can be used by all clinical centres who wish to be part of this international initiative and to share data centrally.
- The representatives of all clinical centres present agreed to share their existing registry/database datasets to facilitate this.
- The Irish Skin Foundation will fund Ian McNicoll of OpenEHR to map these datasets and create a master dataset.
- From this, a core dataset will be selected and approved by a representative working group.
- The core dataset needs to be small, in order to facilitate busy clinicians inputting data to the registry.

The best approach to EB classification

The EB classification, developed by Fine et al[1], referred to as the 'onion skin' classification, is considered gold-standard and should be adopted by the registry.

- Dmitri Wall shared a terminology mapping exercise, based on the onion-skin classification, that might be reviewed by the dataset working group to facilitate mapping to other terminologies such as SNOMED, ICD and Orpha codes.
- The use of OpenEHR will allow flexibility to manage different coding systems.

The importance of data protection

The issue of data protection and consent was a strong focus of the discussion.

- There are very established procedures which can be adopted to manage the de-identification of data shared centrally.
- In relation to patient consent, in general, dynamic consent is the preferred approach
- In cases where re-consent is needed to share annonymised data, it will need to be worked out a local level.

Technology to underpin the initiative

- The Irish Skin Foundation has been working with the software company OpenApp on other skin registries. They will fund OpenApp to develop the initial platform for the Irish EB clinical centres, which can then be adopted by other centres who wish to avail of it, helping to keep the costs down.
- The platform will have an intuitive interface, permit different levels of access and will allow the possibility of a patient portal.

[1]Fine et al., Inherited epidermolysis bullosa: updated recommendations on diagnosis and classification. J Am Acad Dermatol. 2014 Jun; 70(6):1103-26

- Importantly, it will be possible for other centres to maintain or develop their own platforms but still be part of the initiative, should they wish. The use of a core data set and OpenEHR will facilitate this.

Supporting the ERN-Skin

- Any efforts made should be in close collaboration with the proposed ERN-Skin and in harmony with wider EU initiatives.
- The eHealth group of the ERN would benefit from the inclusion of further members from this EB group and this possibility will be investigated.
- There is a planned EU funding call to support patient registries for approved ERNs and the group will aim to apply for this.

The need for governance

It will be important to establish a governance group to oversee the planning and implementation of the registry

- An open invitation should be extended to members of this group to create a registry governance group.
- It is critical that there is strong patient representation on this group, through DEBRA International.
- It was suggested that a scientific committee might also be established to assess the validity of research projects seeking to utilise data from an international EB registry. The governance group (or a subgroup thereof) could also adopt this role.

Conclusion

While this meeting identified numerous obstacles to the establishment of an international EB registry, it also highlighted the willingness of the community to work together towards establishing a network of collaborative EB registries. We believe that, with the resources available to the group and the expertise of its members, it will be possible to develop a sustainable solution that will support existing registries and develop feasible options for new registries. In the context of an international push to develop ERNs, there is a time-sensitive opportunity to develop a bottom-up model registry and ERN platform solution that supports clinical and patient groups, ensuring that information drives better clinical care, patient-involvement and research.