

Submission to consultation on the successor of the Strategy for Science Technology and Innovation

On behalf of people living with epidermolysis bullosa (EB)

SSTI 2006-2013 has delivered measurable impact in terms of the quality of science carried out, increases in human capital and economic benefit (directly and indirectly), improved collaboration between teaching and learning institutes and industry and improved co-ordination of research activities. This was particularly impressive given the huge changes in Ireland's economic circumstances.

Such outcomes demonstrate the positive and tangible impact that such a strategy can have and illustrate why it is so important that we get the next strategy right.

This submission relates to health-related research and its importance to Irish patients, including rare disease patients and, especially, epidermolysis bullosa (EB) patients.

The importance of supporting clinical research

In the Consultation paper, one aspect of STI that receives very little attention is clinical research, despite it being clear that the policies to be delivered by the next SSTI are for both economic and *societal* benefit. This is a significant gap in the Consultation document. We strongly welcome the recent focus on translational health research but advances in translational research can only be realised through testing in the clinic.

Why is clinical research important?

- It is the endpoint of much basic and applied biomedical research in therapeutics.
- If Ireland is to attract the very high value drug discovery and clinical trial activities of the pharma and med tech multinational corporations, it needs to have a strong, functioning clinical research ecosystem
- Equally, indigenous drug discovery and medical device companies need access to clinical research for development and clinical trial activities.
- The undertaking of clinical research within health services has been shown to improve all aspects of patient care.
- Early access to innovative medical interventions will improve the prognosis for Irish patients and increase their likelihood of ongoing access to those treatments.

Suggestions for addressing this issue:

- Make the Health Research Plan part of the SSTI.
- Set human capital targets for research clinicians e.g. numbers trained in good clinical practice etc.
- Align and co-ordinate hospital consultant disciplines with biomedical research strengths.
- Expand the remit of Knowledge Transfer Ireland to include clinicians and hospitals.
- Support and expand disease-specific clinical trial portals i.e. in the manner of the model so successfully demonstrated by ICORG.
- Appoint a research lead in each of the new Hospital Groups (the current post of Chief Academic Officer appears to be primarily focused on training).

The need for public and patient involvement (PPI) in health research

There have been significant advances across Europe and the US in recent years, in involving the public and patients in health research processes. PPI takes many forms, from informing the public about developments in research, to involving patients in research funding decision-making processes, to patient organisations leading a research agenda. PPI was the topic of the first forum of the Irish Health Research Forum (<http://www.ihrf.ie/>). See the meeting report here: <http://bit.ly/1N88NFV>.

Why is PPI important?

- Informing patients about research that is relevant to them is likely to result in their increased support for research and the improved likelihood of patient participation in clinical trials (patient participation in clinical trials is increasingly becoming a barrier to the progression of medical research).
- PPI gives patients and their representatives the opportunity to express the questions and needs that matter most to them, thereby improving the relevance of research.
- PPI helps to ensure that clinical research studies are designed to be sensitive to the needs of the subjects participating in them.

Suggestions for addressing this issue:

- Including reference to the importance of PPI in the SSTI will help to ensure that it becomes part of the vocabulary of health research in Ireland.
- Initial steps by the HRB to implement PPI should be supported and expanded.
- All health research funding and policy agencies should take steps to formally implement PPI in their processes.
- In implementing the strategy, patient organisations should be routinely consulted in all aspects of State health research policy.

The requirement to support the entire pipeline of health research

Concerns by the scientific community, in relation to the balance of funding between basic and applied research, have been given considerable attention recently. We will not re-iterate those often-voiced concerns but wish to emphasise that, as patient representatives, we stand firmly beside the 1000+ Irish scientists who have recently voiced their disquiet, through the medium of a letter to the Irish Times. Though we believe that very little research in the realm of health falls into the category of basic research, early stage bio-medical research is of particular importance to rare diseases, where knowledge of the disease is often limited. It is time to renew our focus on excellence and reward the pockets of strong rare disease research, where they exist.

Suggestions for addressing this issue:

- Health research should be funded, first and foremost, on the basis of excellence.
- While we understand that the Irish people have a desire to see a reduction in unemployment figures, they equally wish to know that they can depend on their health service. We believe that health research should not be subject to the same need to demonstrate short-term economic impact as other areas of research.
- It is imperative that the various State funding agencies with a health research remit take a co-ordinated approach to funding. Each agency currently has a strategy which is based on strong logic but which has been developed in isolation. A coordinated approach to funding health research is essential, in order to avoid the detrimental research funding gaps which have emerged in the system.
- The fact that policy decisions in relation to research have, in recent times, been driven by the Department of Jobs Enterprise and Employment, has led to an imbalance in the decision-making focus around research. We

would like to see the Department of Health play a more prominent role in policy decisions around health research.

- The creation of a database of Irish research and researchers (akin to the planned HEA Large Items of Research Equipment Database) would play a valuable role in improving research success. It would support Irish researchers in developing the research networks which are a pre-requisite for seeking Horizon 2020 funding. This would be of particular value in the field of rare disease research, in which researchers are often isolated. Such a network is called for in the recently published National Rare Disease Plan for Ireland: <http://health.gov.ie/wp-content/uploads/2014/07/EditedFile.pdf>

The need for high quality health information

With the lack of unique health identifiers in Ireland to date and insufficient support to develop high quality and sustainable registries, Ireland has very poor information on which to base its health services.

Why are patient registries important?

High quality patient registries, which can share data with other registries internationally, are of particular importance in the case of rare diseases for many reasons, including the following:

- They support a more streamlined and connected approach to clinical care.
- They allow the identification of patients for clinical trials.
- They improve the understanding of the natural history of a disease (an essential prerequisite to determining whether or not an intervention has had a positive effect).
- They permit post-marketing analysis of a drug or intervention.

Suggestions for addressing this issue

- There is currently no Irish funding source for patient registries, as the financing of such infrastructure does not fit within the remit of State funding agencies. Changing this however would have numerous benefits for the field of health research as a whole.
- While the appointment of Chief Information Officer within the HSE, to implement individual health identifiers within the health service, is welcome, it is important to ensure that this position is sufficiently resourced to fulfil its role in the near term.

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DEBRA Ireland is the patient support organisation for people who live with EB (epidermolysis bullosa). We provide patient support services, drive research into treatments and cures and advocate for those living with EB.

Guide to EB

What is EB?

Epidermolysis bullosa (EB) refers to a group of distressing and painful genetic conditions that cause the layers of the skin and internal mucous membranes to separate and blister at the slightest touch.

How is EB caused?

EB is a genetic condition and is inherited in either a dominant or a recessive fashion. It is chronic and debilitating.

How common is EB?

EB is very rare. An estimated 1 in 18,000 babies born are affected by it.

What genes cause EB?

There are a number of genes associated with the structure of skin, that can cause EB if they have a genetic fault in them. These include genes coding for collagen, keratin and integrin proteins.

Are there different types of EB?

Yes, there are different forms of EB which vary in severity. Broadly, there are three major subtypes: EB simplex, junctional EB and dystrophic EB. EB simplex tends to be a less severe form of the condition (although still very debilitating) but there are exceptions and some subtypes of EB simplex are very severe. Babies born with junctional EB rarely survive. Dystrophic EB ranges in severity but usually has a huge impact on quality of life and, at the severe end of the scale, is a truly devastating condition. Severe forms are progressive, due to, among other things, the build-up of scar tissue.

How is EB diagnosed?

If EB is suspected, a biopsy is usually taken. Microscopy and antigen mapping are used to guide the diagnosis. In Ireland, the diagnosis is almost always confirmed by genetic testing.

Is it only the skin that is affected in EB?

In less severe cases, the symptoms of EB are localised to the skin. In more severe cases the mucous membranes (mouth, oesophagus and anal tract) are often badly affected, along with many other parts of

the body, including teeth, nails, eyes and bones. In severe cases there is almost no part of the body that remains unaffected and the care of EB requires a large multidisciplinary team of medical specialists.

Are there effective treatments for EB?

No, there are no effective treatments for EB. Current care involves the management of symptoms, which has improved dramatically over the years. There is much research being undertaken world-wide however, into developing effective treatments.

How are EB patients cared for in Ireland?

EB patients are cared for by specialist, multidisciplinary teams in Our Lady's Children's Hospital Crumlin and St. James's Hospital. This care is supplemented by support from a variety of specialists in the primary care setting, including GPs, community nurses, OTs, physiotherapists, dentists etc.

What financial costs are associated with EB?

There are a variety of costs associated with EB. The largest expense is usually that associated with dressings and bandages which, in severe cases, can run to several thousand euro a month. There are also many medications involved in the care of EB including pain relievers, anti-itch medications, nutritional supplements, anti-depressants, antibiotics, laxatives, ophthalmic treatments and emollients. Finally there is often a broader financial impact on the family, relating to loss of earnings and travel to specialist care providers.

EB Clinical Images

