EUROPEAN COMMISSION
DIRECTORATE-GENERAL FOR HEALTH AND FOOD SAFETY

Public health, country knowledge, crisis management
Health programme and chronic diseases

Luxembourg, 28th February 2019

MISSION REPORT

MIPS NO	**DF-19-1676863**
CHARGÉ DE MISSION	
UNIT	**SANTE C1**
TO	**BRUSSELS**
WHEN	**20 FEBRUARY 2019**
SUBJECT	**LAUNCH OF THE 'GLOBAL COMMISSION' RECOMMENDATIONS ON RAPID DIAGNOSIS OF RARE DISEASES**
COUNTERPART(S)	**MICROSOFT, TAKEDA, EURORDIS**
OBJECTIVE OF THE MISSION:	**TO REPRESENT DG SANTE IN A PANEL DISCUSSION**

MAIN ISSUES DISCUSSED AND WHAT HAS BEEN ACHIEVED IN VIEW OF THE OBJECTIVE

Shire (Now Takeda), Microsoft and EURORDIS set up a 'Global Commission' to accelerate time to diagnosis for children with rare diseases. The Commission is jointly led by (Eurordis), Microsoft, and According to its web site:

'The Global Commission to End the Diagnostic Odyssey for Children ("the Global Commission") is a multi-disciplinary group of experts with the creativity, technological expertise and commitment required to make a major difference in the lives of millions of children and their families. The Global Commission will develop an actionable roadmap to help the rare disease field to shorten the multi-year diagnostic journey, considered a key to a longer, healthier life.

Within its roadmap, the Global Commission will offer recommendations designed to address core barriers preventing timely diagnosis impacting all rare disease patients, of which approximately half are children, such as:

• Improving physicians' ability to identify and diagnose patients with a rare disease in order to begin care and treatment

• Empowering patients and their families to have a more active role in their health care

Commission européenne, 2920 Luxembourg, LUXEMBOURG - Tel. +352
Office: HTC - Tel. direct line +352 - Fax +352 4

@ec.europa.eu

• Providing high-level policy guidance to help achieve better health outcomes for rare disease patients'

At the meeting, which took place simultaneously in Brussels and New York by videolink, the recommendations of the 'Global Commission' were published, in 3 areas ('tracks'):

Patient and Family Empowerment

Create or identify new tools and approaches to empower patients and caregivers so they can navigate the health system more effectively

Equipping first-line providers with tools for diagnosis and referral

Apply innovation and creative thinking to improve primary care physicians' ability to identify patients with a rare disease and refer to appropriate follow-up care

Reimagining the Genetic Consultation

Develop innovative ways to enable geneticists and specialists to operate more efficiently so they can see more rare disease patients quicker – especially given the growing shortage of geneticists

The specific recommendations (published only in the form of powerpoint slides) were:

- Empowering Patients to Ask their Doctor to Think Differently
- Portable Blockchain Health Records
- Use Artificial Intelligence to Identify Rare Diseases
- Facilitate and Expand Access to Diagnostic Testing
- Establish Early-Access Centers in Genetics Clinics
- Information Capture and Tele-Consultations for Rural/ Remote Patients

In my intervention (a 3 minutes slot!) I referred to key aspects of EU work on rare diseases, much of which is directly related to the recommendations of the 'Global Commission'. I mentioned IRDIRC, with the same objective of shortening waiting times for diagnosis, about 50 partners from five continents already working since 10 years, and major breakthroughs already achieved. I referred to the ERNs, which offer many services to patients also envisaged by the 'Global Commission'. I highlighted the launch of the rare disease registration platform by the JRC on 28 February.

POSSIBLE OTHER CONTACTS IN MARGIN OF THE EVENT

I invited [redacted] Health and Life Sciences EMEA, Microsoft, to contact us if there is any interest to co-ordinate this initiative with our activities on rare diseases (which I explained [redacted] in more detail during a coffee break).

FOLLOW UP TO BE GIVEN

To see if there can be synergies between this initiative and the many activities of the

Commission in the area of rare diseases.

OTHER REMARKS

The ‘Global Commission’ created by Microsoft, Takeda, and Eurordis leaves many questions open. Instead of taking action, recommendations are issued, of which we have already too many. The recommendations are supposed to be ‘actionable’, but are at a very high level, so that real implementation remains doubtful, in particular because it is unclear to whom they are addressed. It is also unclear if these recommendations would not increase health inequalities, because they might tend to improve options mainly for well educated people.

At the same time this initiative has the potential to draw away attention from the implementation of recommendations that exist already. So what exactly is the added value (other than having Microsoft on board?)