Inspirational Speech at the European Conference on Rare Diseases

Dr Rüdiger Krech, Director, Universal Health Coverage and Health Systems, Office of the Assistant Director-General, World Health Organization

Vienna, 11 May 2018

Your Excellencies, dear Mr Seychell, dear Yann and Paloma,

"The vision of the Sustainable Development Goals is a world in which no one is left behind, including people who suffer from rare diseases. Just because a disease affects a small number of people does not make it irrelevant or less important than diseases that affect millions." This is a quote that shall be the basis of my speech today. It comes from WHO's Director-General, Dr Tedros.

With 300 - 350 million people affected worldwide, and more than 7,000 different types of diseases known to date, rare diseases represent a major challenge in public health that has been largely ignored. Consequently, this is a field in public health and research that would certainly benefit from globally concerted action and international collaboration.

Why, we may ask, are rare diseases largely ignored? While I will not even try to give a comprehensive answer, I'd rather want to highlight some dimensions and offer some questions that might inspire our debate.

Konrad Lorenz, one of the founding fathers of the field of ethology, has said that "every researcher gets a narrower and narrower field of knowledge in which he (and my I add: she) must be an expert in order to compete with other people. The specialist knows more and more about less and less."

As the body of scientific knowledge in a discipline increases, there is pressure for specialization.

Fields spawn subfields that then become entities in themselves that promote further specialization.

Scientific specialisation has been hugely successful in the area of identifying rare diseases. But it has also led to isolation as well as poor visibility and public attention.

Viewing Rare Diseases through a Global Public Health lens offers a different perspective:

Rare disease patients and their families are a particularly vulnerable group of citizens who experience scarcity of medical knowledge, difficulties in accessing adequate care, as well as isolation from society due to the rarity of their condition and the scattered expertise.

In general, health systems are not adapted to rare diseases and there is little public health policy to respond to their specific needs: access to quality health care, overall social and medical support, effective liaison between hospitals and general practices, as well as professional and social integration, autonomy and independence.

Universal Health Coverage, means that all people and communities receive the promotive, preventive, curative, rehabilitative and palliative health services they need, of sufficient quality to be effective, while also ensuring that the use of these services does not expose the user to financial hardship.

Health systems can learn a lot from Rare Diseases to design systems that are fit for the future and effectively contributing to UHC. Rarity calls for increased international mobility of experts, as well as of patients, as the most clinically relevant expertise will most likely not be available locally. Will innovative solutions in e- and m-health be able to pool expertise and develop virtual Centres of Expertise? Could they help to create expert structures for the management and care of rare disease patients and bring together multidisciplinary competences and skills, in order to serve the specific medical, rehabilitative and palliative needs of rare diseases patients? Could accredited global centres of excellence help to provide best quality care based on available clinical evidence? Would we be able to globally find innovative ways of financing such centres so that patients and their families will not fall into poverty because of Out of Pocket payments related to the care of their Rare Disease?

“Pooling” seems to be one of the magic words when it comes to Rare Diseases. It helps to bring together the critical mass and balance low prevalence and incidence. It helps to focus the diffuse shadow light in which Rare Diseases are currently barely seen and create spot lights: pooling of Research and Development; pooling of data; pooling of care facilities and pooling of experts. Lots of excellent work has been undertaken in recent years in this regard. The launching of the European Reference Networks is one of such initiatives. Could “pooling” also serve to bring in expertise from Japan, Australia, Canada or China?

A second word that comes to mind is “collaboration”. Sean Ekins and colleagues reviewing the recent Charcot-Marie-Tooth research and priorities suggest that “it is unlikely that we are going to see a dramatic change unless there is a wholesale shift in the process of drug discovery and development, combined with increased collaboration between academics, industry, government labs and research foundations in the rare disease arena.” How will such a shift be concretely

orchestrated? What incentives could be created that instigate such change?

One such incentive could be to collaborate to agree on thresholds for safe and clinically effective standards for diagnosis, care and treatment, and to collect international best practice.

And a third word might be “smart innovation”. As technologies are advancing, we are confident that we will see routine in vivo gene editing in a medium time horizon. I added “smart” to “innovation”, as again, the global community can and certainly will learn from Rare Diseases: we are faced with the option of editing the human genome with ease and precision. Notwithstanding the potential of these applications to be beneficial to any one individual, unforeseen risks such as epigenetic and inter-generational effects cannot be ruled out. Besides principled reasons, it is also because of these uncertainties, that interventions seeking to modify the human genome are banned in many countries. On the other hand, the question posed by the US National Academy of Sciences and National Academy of Medicine is no longer *whether* or not the human genome ought to be edited – rather the question is *what* should be the criteria for heritable germline editing, and what principles should guide the governance of human genome editing. Such a shift in the approach to germline editing would probably have global consequences for individuals, societies and mankind altogether.

So, on the way towards “cure” of some Rare Diseases, I humbly suggest to go this path with caution.

Colleagues, I am sure that pooling, cooperation and smart innovation will help to increase the attention to Rare Diseases globally. WE are looking forward to being at your side.

Thank you.