House Committee on Appropriations: Subcommittee on Labor, Health and Human Services, Education and Related Agencies
Public Witness Testimony

Testimony of Brett Kopelan, Executive Director, debra of America

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I thank Chairman Cole, Ranking Member DeLauro, and Members of the Subcommittee for providing this opportunity to submit Public Witness written testimony to the Subcommittee on matters of importance to the Dystrophic Epidermolysis Bullosa Research Association of America (debra of America, www.debra.org) with respect to activities within the National Institutes of Health (NIH).

My name is Brett Kopelan, and I serve as the Executive Director of debra of America, which is the only U.S. nonprofit providing all-inclusive support to the Epidermolysis Bullosa (EB) community. We accomplish our mission to increase the quality of life for all those living with EB by doing two things in parallel; funding the most innovative science intended to treat or cure EB, and providing free programs and services for those living with EB. Throughout its almost 40 year history, debra of America has remained committed to our mission to serve the EB community in this capacity.

EB is a rare genetic connective tissue disorder with many genetic and symptomatic variations, yet all share the prominent symptom of extremely fragile skin that blisters or tears from minor friction or trauma. EB occurs in 1 out of every 20,000 births in the United States

debra of America ϕ 75 Broad Street, Suite 300 ϕ New York, NY 10004 ϕ (P) 212-868-1573 ϕ (F) 212-868-9296 ϕ debra.org

(approximately 200 children a year are born with EB), affects both genders as well as every racial and ethnic group equally. Severity of the disease runs the gamut from mild to lethal in the first few months of life. EB has been called a skin disease but that is reductive. Internal organs and bodily systems can be severely affected. In the more severe forms, there is a litany of secondary illnesses and complications that require multiple interventions from a variety of medical specialties. As of today, there is no FDA approved treatment. Pain management, wound care, and preventative bandaging are the only treatment options available. The specialized wound care products can cost a family in excess of one million dollars a year.

My proudest title I hold is father to Rafaella Lily Kopelan, a 10-year-old girl with Recessive Dystrophic EB. Rafi was born in October of 2007 and diagnosed with a severe form of EB ten days after birth. Fifteen minutes of online research scared me to the bone and broke my heart into a million pieces. While there was ongoing academic research, nothing could be considered translational and there was no biotechnology or pharmaceutical activity or interest. With no U.S. Food & Drug Administration (FDA) approved treatment and no cure on the horizon, my worst nightmare as a new father was realized. But I was wrong. Rafi is one of the most heroic and incredible children on the planet, yet watching her live daily with EB and trying to manage her symptoms is a never-ending nightmare where everyday a little piece of my heart breaks still.

Rafi is one of approximately 25,000 Americans and 500,000 patients worldwide living with EB. The resilience and courage of her and others living with EB is nothing short of heroic given the pain, suffering, trauma, and overall general challenges of daily life. The plight of EB is the reason we refer to it as “the worse disease you’ve never heard of”. Similar to most of the 7,000 FDA recognized rare diseases, many in the medical field have never seen a patient with EB

meaning they have no practical or clinical experience treating the disease. With such a small patient population, natural challenges in terms of basic awareness of EB in the medical community, research community, and, in general, the public at large is common place.

The good news is that the lack of institutional and public awareness tide is turning. At the federal government level, thanks to your Subcommittee's leadership and others in the United States' Senate through Committee report language and research support, EB is starting to become a more familiar name and gain traction in Congress and at the National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS) at the NIH. Industry partners have begun to invest capital and develop drugs and therapies while clinicians throughout the country are becoming better versed and more equipped to treat EB patients. For the first time, I can see light at the end of a tunnel and know it's not an oncoming train. Hope and an expectation of a better quality of life for my daughter and others living with EB is replacing despair. We are on the forefront of gene therapy, gene editing, as well as other regenerative medicine initiatives that one day in the near future will provide relief for those with EB and potentially open new doors for therapies for other diseases.

Our efforts in hosting annual International Research Consortiums, where we convene the world's premiere academic researchers, and biotechnology sponsors to provide updates to ongoing initiatives while providing an opportunity to plan collaborative studies is something we are proud to lead. Furthermore, we also host a biennial debra Care Conference for EB families, caregivers, clinicians, researchers, and related medical suppliers. While we understand the responsibility to help address the practical needs of our EB community, we also recognize that a parallel effort

educating, informing, and advocating for assistance and tools from our elected leaders in Congress is necessary.

There is more that can be and must be done by our federal officials to help this vulnerable population. Public and private partnerships are the way forward. We encourage leaders at the NIH to convene a national coordinating committee on EB, including representatives of relevant federal agencies and members of the EB community, to address any gaps in EB educational and research efforts. Such an interagency committee could be empowered to build from the successful models of other disease states (such as muscular dystrophy), which has led to efficiencies and the development of centers of excellence as a focal point for basic and translational research support. A national surveillance epidemiology and disease monitoring program at Centers for Disease Control (CDC) to collect data about the treatment strategies for EB would advance many productive opportunities to benefit the EB population. Better understanding of current treatment paradigms at the EB population-level has the potential to improve patient health care outcomes and obtain valuable new information about important public health factors, such as emerging antimicrobial resistant pathogens. The empowerment of a new generation of life sciences researchers, health care providers, and other aligned public health professionals through federal research support to obtain accurate information regarding the nature, effects, disease management, and newly available techniques of treatment options for EB could be very meaningful for not only our patient population but also other rare diseases. Interagency data sharing, and direct involvement by community members, including medical and research professionals active in the field, can have a catalyst effect upon the private sector. We also believe that the Health and Medicine Division of the National Academies, formerly the Institute of Medicine (IOM), should

organize an EB consensus conference to address gaps and identify consensus best practices for current standards for care provision, educational and social supports, and other special topics particular to the support of well-being for persons with EB.

Mr. Chairman, Ranking Member, and Members of the Subcommittee, we greatly appreciate your attention to the needs of the EB and rare disease communities and to the importance of research towards advancing significant therapies and cures for the American people. We desire to be the trusted resource to Congress, the Administration, and relevant federal agencies in the EB and rare disease enterprise.

Thank you for your leadership in the United States Congress and your willingness to receive our written testimony. debra of America, and the entire EB community applaud your service to our nation and for the attention you bring to the plight of the ten percent of Americans —including my daughter Rafi –who suffer from rare diseases.

debra of America stands ready to work alongside you in these important efforts. Thank you.