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783rd Conference

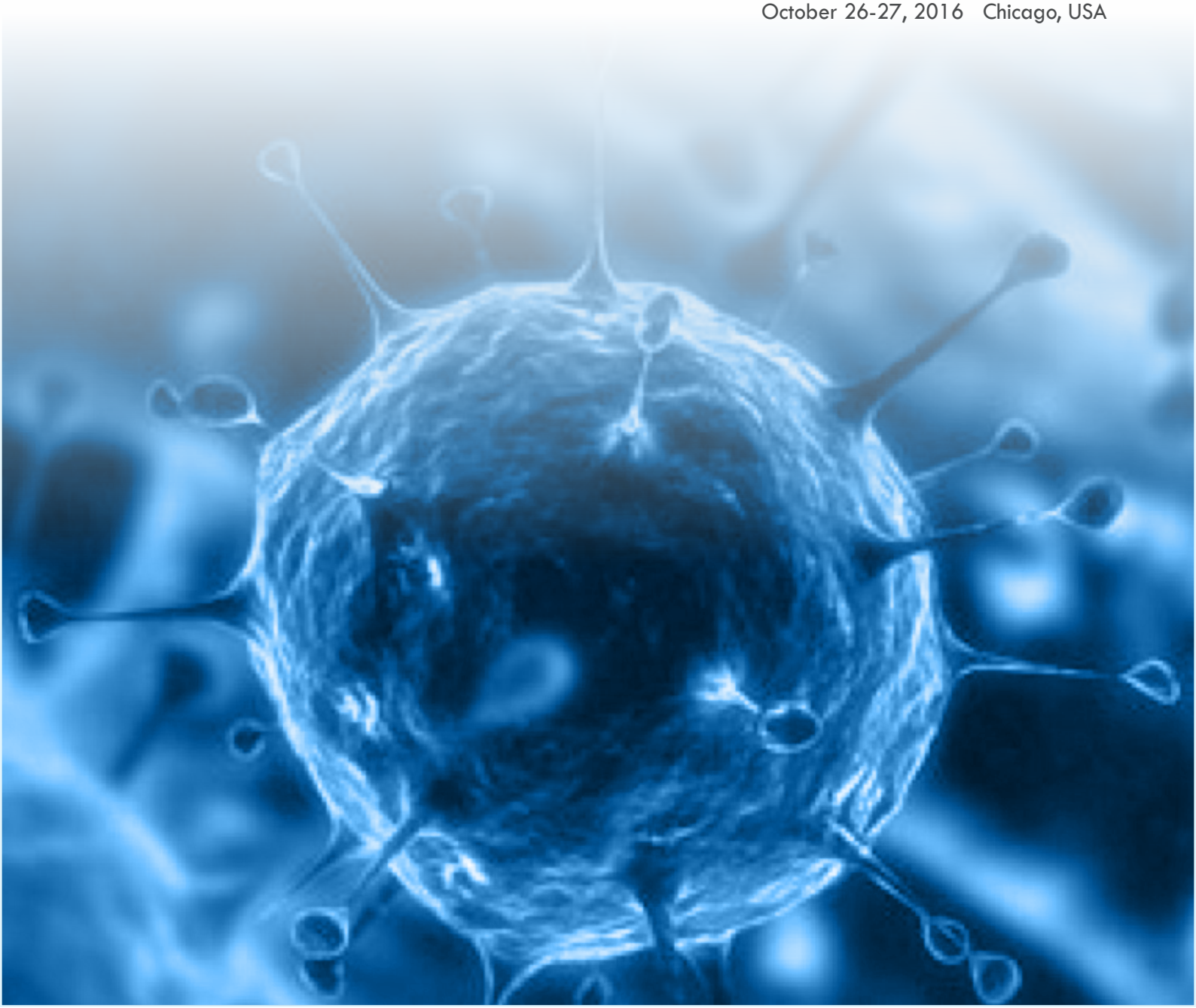
Scientific Program

Proceedings of

Annual Congress on

RARE DISEASES AND ORPHAN DRUGS

October 26-27, 2016 Chicago, USA



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Day 1 October 26, 2016

08:00-09:00 Registrations

Hall

conferenceseries.com 09:00-09:25 **Opening Ceremony**

Keynote Forum

Introduction

09:25-10:10 **Title: The rare diseases clinical research network (RDCRN) program (a model for collaborative research) and resources at NCATS, NIH**

Rashmi Gopal-Srivastava, NCATS, NIH, USA

10:10-10:55 **Title: Inside track to an orphan designation**

Timothy Coté, Coté Orphan, USA

Networking and Refreshment Break 10:55-11:15

Group Photo

Major Sessions:
Different types of Rare Diseases | Clinical Research and Public Awareness | Mystery Diagnosis of Rare Diseases

Session Chair: Amrik Sahota, Rutgers University, USA
Session Co-chair: Lisa Baumbach- Reardon, Translational Genomics Research Institute, USA

Session Introduction

11:15-11:35 **Title: Tailored inhibition of cystine stone formation as a therapy for cystinuria**
Amrik Sahota, Rutgers University, USA

11:35-11:55 **Title: Failures in brain energy metabolism unveil therapeutic targets for Huntington`s disease**
Maite A. Castro, Universidad Austral de Chile, Valdivia, CHILE

11:55-12:15 **Title: Epilepsy & rescue meds in schools**
Tyson E. Dewsnup, Epilepsy Association of Utah, USA

12:15-12:35 **Title: Neo212: A new drug for temozolomide resistant malignant gliomas**
Thomas C Chen, Keck School of Medicine of USC, USA

12:35-12:55 **Title: Management of life threatening hyperammonemia in children**
Philippe Jouvet, Sainte-Justine Hospital University of Montreal, Canada

Lunch Break 12:55-13:40

13:40-14:00 **Title: Defending the Dream; perils beyond science and finance**
Laura Sunderlin, Beazley, USA

14:00-14:20 **Title: Engaging families in research to drive progress: The phelan-mcdermid syndrome international registry (PMSIR) and the phelan-mcdermid foundation data network (PMS_DN)**
Megan O'Boyle, Phelan-McDermid Syndrome Foundation, USA

14:20-14:40 **Title: Fever of unknown origin: Case study**
Yusuf Hovsep Eken, Elkerliek ziekenhuis, Netherland

14:40-15:00 **Title: A 20 days old patient with genital asymmetry**
Marise Abdou, Abo El-Rish Children's Hospital, Cairo, Egypt

15:00-15:20 **Title: Association between fbn1 polymorphisms and tgf- ß1 concentration within aneurysms and dissections of ascending thoracic aorta**
Ramune Sepetiene, Lithuanian University of Health Sciences, Kaunas, Lithuania

Major Sessions:
Challenges in Rare Diseases Treatment |Rare Diseases in Cancer | Rare Infectious Diseases and Immune Deficiencies

Session Chair: Lisa Baumbach- Reardon, Translational Genomics Research Institute, USA

15:20-15:40 **Title: On-going exon 53 skipping clinical trial for Duchenne Muscular Dystrophy**
Shin'ichi TAKEDA, National Institute of Neuroscience, Japan

15:40-16:00 **Title: Rare disease research: Opportunities and challenges**
Lisa Baumbach- Reardon, Translational Genomics Research Institute, USA

Networking and Refreshment Break 16:00-16:20

16:20-16:40 **Title: COST Action BM1207: Involving all stakeholders to overcome challenges of genetic therapy development for Duchenne Muscular Dystrophy**
Annemieke Aartsma-Rus, Leiden University Medical Center, The Netherlands

16:40-17:00	**Title: Overlap of metabolic and endocrine dysregulation during orphan disease-special focus on cardiovascular disease** **Prasanth Puthanveetil** , Roosevelt University College of Pharmacy, USA
17:00-17:20	**Title: Tapping untapped: Exploring role of ALDH in pharmacogenetic and toxicogenetic studies** **Nasir Ali Afsar,** Alfaisal University College of Medicine, Riyadh, KSA

Panel Discussion

Day 2 October 27, 2016

Hall

Keynote Forum

09:35-10:20 **Title: How patient organizations can change the game in innovation and drug marketing: Example of AFMTelethon**

Serge Braun, Scientific Director, AFMTelethon, France

Major Sessions:
Orphan Drugs, Development Trends and Strategies | Orphan Drugs and Ethical Issues | Future Hereditary and Rare Diseases e Prospects of Rare Diseases

Session Chair: Alan B. Moy , The John Paul II Medical Research Institute and Cellular Engineering Technologies, USA

Session Introduction

10:20-10:40	**Title: Network pharmacology based repurposed drugs combination for orphan diseases treatment** **Daniel Cohen,** Pharnext , France

Networking and Refreshment Break 10:40-11:00

11:00-11:20	**Title: JBPOS0101: A new generation mGluR- and BBB-targeted broad spectrum antiepileptic drug for the treatment of super-refractory status epilepticus** **Yong Moon Choi,** Bio-Pharm Solutions Co., Ltd, South Korea
11:20-12:00	**Title: Cellular therapies for rare disease** **Stephen Shrewsbury,** Fortuna Fix, USA
12:00-12:20	**Title: Digital measurements of health- regulatory science challenges opportunities in rare diseases** **Stephen P. Arneric,** Critical Path Institute, Tucson, AZ, USA
12:20-12:40	**Title: Novel viral-free and oncogene-free induced pluripotent stem cell for orphan disease cell therapies** **Alan B. Moy,** The John Paul II Medical Research Institute and Cellular Engineering Technologies, USA
12:40-13:00	**Title: Orphan drugs: Getting arms around rare diseases** **Duygu Kuyuncu Irmak,** INC Research, Turkey

Lunch Break 13:00-13:40

13:40-14:20	**Title: Pricing, public health, & politics: Policy considerations for orphan products** **Anne Marie Finley,** Biotech Policy Group LLC, USA
14:20-14:40	**Title: The role of clinical genomic testing in treatment discovery for rare neurodevelopmental diseases** **Karen S. Ho,** Lineagen, Inc, USA
14:40-15:00	**Title: Rare disease studies facilitated by taking study visits to the patients** **Gail Adinamis,** GlobalCare Clinical Trials, LLC, USA
15:00-15:20	**Title: A robust reprogramming approach to create viral-free and oncogene-free, orphan-disease specific induced pluripotent stem cells from peripheral blood mononuclear cells** **Alan B. Moy,** The John Paul II Medical Research Institute and Cellular Engineering Technologies, USA
15:20-15:40	**Title: Challenges and opportunities in development of orphan drugs** **Moji C. Adeyeye,** Roosevelt University, USA
15:40-16:00	**Title: Potential treatments for rare diseases:Cell therapy, gene therapy and genome editing** **Jacques P. Tremblay,** Department of Molecular Medecine, Université Laval, Québec, Canada

Networking and Refreshment Break 16:00-16:15

Poster Presentations

RD-01	**Title: An online survey of neurologists about charcot-marie-tooth disease type 1A** **Xavier Paoli,** Pharnext, France
RD-02	**Title: The first case report of *Raoultella planticola* liver abscess** **Moamen Al Zoubi,** Advocate Illinois Masonic Medical Center, Chicago, USA
RD-03	**Title: Rapid progression osteolysis in Gorham Stout Syndrome: A case report and literature review** **Leonardo Cano,** Universidad Católica Santiago de Guayaquil, Ecuador

Award Ceremony