WORLD*Symposium*™ 2022 Preliminary Program*
Platform Presenters

Monday, February 7, 2022
Basic Science

7:30	Chester B. Whitley University of Minnesota Minneapolis, MN, United States	Welcome & Announcements Presentation of 2022 Roscoe O. Brady Award for Innovation and Accomplishment to Stuart A. Kornfeld
7:35	Stuart A. Kornfeld Washington University St. Louis, MO, United States	Innovation Award Speaker Presentation
8:00	Allisandra Rha CHOC Children's Research Institute Orange, CA, United States	Prime editing corrects the c.1826dupA mutation in infantile-onset Pompe disease
	Travis Moore Sainte-Justine Research Center Montreal, QC, Canada	IPSC derived neurons of mucopolysaccharidosis type III patients show pronounced synaptic defects **2022 Young Investigator Award Recipient*
	Oriana Mandolfo The University of Manchester Manchester, United Kingdom	Systemic immune challenges exacerbate inflammation and cognitive decline in a mouse model of MPS IIIA **2022 Young Investigator Award Recipient*
	Mahsa Taherzadeh McGill University Montreal, QC, Canada	Expression of misfolded HGSNAT protein aggravates neurological phenotype in mucopolysaccharidosis type IIIC **2022 Young Investigator Award Recipient*
	Live Moderated Q&A	*Allisandra Rha, Travis Moore, Oriana Mandolfo, and Mahsa Taherzadeh*
9:00	Miles Smith University of Minnesota Minneapolis, MN, United States	Comparative effectiveness of intravenous and intrathecal AAV9.CB7.hIDS (RGX-121) in a murine model of mucopolysaccharidosis type II
	Gani Perez NHGRI, National Institutes of Health Bethesda, MD, United States	Behavioral and whole transcriptome analyses of a *gba*-haploinsufficient Parkinson murine model
	Tsui-Fen Chou California Institute of Technology Pasadena, CA, United States	Enzyme replacement therapy (ERT) for MPS IIID
	Marya Sabir National Human Genome Research Institute, National Institutes of Health Bethesda, MD, United States	A novel experimental mouse model to investigate a free sialic acid storage disorder (Salla disease) **2022 Young Investigator Award Recipient*
	Live Moderated Q&A	*Miles Smith, Gani Perez, Tsui-Fen Chou, and Marya Sabir*
10:00	Break	

10:30	Elizabeth Braunlin University of Minnesota Minneapolis, MN, United States	Aortic dilation in murine mucopolysaccharidosis type I: A tale of two strains
	Ikhui Kho McGill University Montreal, QC, Canada	Severe kidney dysfunction in the mouse model of sialidosis reveals novel role of neuraminidase 1 in reabsorption process **2022 Young Investigator Award Recipient*
	Fiona Weaver McMaster University Hamilton, ON, Canada	Endoplasmic reticulum stress derives neurodegeneration in the spinal cord of Sandhoff disease mice **2022 Young Investigator Award Recipient*
	Sarah Kim University of Minnesota Minneapolis, MN, United States	Cerebrospinal fluid chitotriosidase as a surrogate endpoint of the efficacy of the PS gene editing system in neurodegenerative lysosomal diseases
	Live Moderated Q&A	*Elizabeth Braunlin, Ikhui Kho, Fiona Weaver, and Sarah Kim*
11:30	Break and Satellite Symposia	
1:00	Gisele Pino Mayo Clinic Rochester, MN, United States	The synergy of multiplex testing to screen for lysosomal disorders (LD)
	Xuefang Pan CHU Ste-Justine Research Centre, Université de Montreal Montreal, QC, Canada	Neurodegenerative role of lysosomal cathepsin B in MPS IIIC
	Francyne Kubaski UFRGS/HCPA Porto Alegre, Brazil	Prenatal diagnosis of mucopolysaccharidosis type VI by analysis of the amniotic fluid supernatant in the mass spectrometry era
	Sukirhini Balendran Medical University of Vienna Vienna, Austria	Rapid identification of IOPD and early-onset Pompe disease by biochemical enzymatic testing followed by genetic confirmation
	Live Moderated Q&A	*Gisele Pino, Xuefang Pan, Francyne Kubaski, and Sukirhini Balendran*
2:00	Malte Lenders University Hospital Muenster Muenster, Germany	Isolation and characterization of a polyclonal human anti-drug antibody as a reference in Fabry disease
	Sireesha Murala Duke University Medical Center Durham, NC, United States	Diffusion tensor imaging (DTI) findings in children with Pompe disease: Insights into white matter hyperintensities from a longitudinal study **2022 Young Investigator Award Recipient*
	Walla Al-Hertani Boston Children's Hospital Boston, MA, United States	A 3-year pilot screening program for lysosomal disorders in the Latin America (LATAM) region using an integrated enzymatic and molecular approach
	Joseph Muenzer University of North Carolina Chapel Hill Chapel Hill, NC, United States	Fifteen years of the Hunter Outcome Survey (HOS): Real-world insights into the patient population living with mucopolysaccharidosis type II (MPS II)
	Live Moderated Q&A	*Malte Lenders, Sireesha Murala, Walla Al-Hertani, and Joseph Muenzer*
3:00	Poster Session in the Exhibit Hall	
5:30	Satellite Symposia	

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Tuesday February 8, 2022
Translational Research

7:30	Chester B. Whitley University of Minnesota Minneapolis, MN, United States	2022 Patient Advocate Leader (PAL) Award Announcement and Presentation to Sue Kahn
7:45	Chester B. Whitley University of Minnesota Minneapolis, MN, United States	2022 Young Investigator Awards Announcement and Presentation
8:00	Jennifer Cohen Duke University Durham, NC, United States	In utero enzyme replacement therapy in a fetus with infantile-onset Pompe disease
	Tahseen Mozaffar University of California Irvine Irvine, CA, United States	AT845 gene replacement therapy for late onset Pompe disease: Overview of clinical data from FORTIS, a phase 1/2 open-label clinical study
	Kevin Flanigan Nationwide Children's Hospital Columbus, OH, United States	Interim results of Transpher A, a multicentre, single-dose, phase 1/2 clinical trial of ABO-102 investigational gene therapy for Sanfilippo syndrome type A (mucopolysaccharidosis type IIIA)
	Tierra Bobo University of North Carolina at Chapel Hill Chapel Hill, NC, United States	Facilitate by-stander effects by EV-mRNA cargo in AAV gene replacement therapy for treating MPS IIIC **2022 Young Investigator Award Recipient*
	Live Moderated Q&A	*Maximiliano Presa, Lalitha Belur, Stuart Ellison, and Michael Przybilla*
9:00	Maximiliano Presa The Jackson Laboratory Bar Harbor, ME, United States	Efficacy of a scAAV9/*SUMF1* viral vector for the treatment of multiple sulfatase deficiency
	Lalitha Belur University of Minnesota Minneapolis, MN, United States	Treatment of cardiac, neurologic, and skeletal manifestations of murine MPS I with AAV9-IDUA: Efficacy study of vector dose and route of administration
	Stuart Ellison University of Manchester Manchester, United Kingdom	Enhanced transduction and immunophenotyping demonstrates preclinical safety and efficacy of haematopoietic stem cell gene therapy for mucopolysaccharidosis type II using an IDS.ApoEII brain targeted therapy
	Michael Przybilla University of Minnesota Minneapolis, MN, United States	Prevention of murine GM1-gangliosidosis following heterotopic insertion of *Glb1* using gene editing
	Live Moderated Q&A	*Maximiliano Presa, Lalitha Belur, Stuart Ellison, and Michael Przybilla*
10:00	Break & Exhibits	

Tuesday February 8, 2022
Translational Research *(cont.)*

10:30	Jonathan Cooper Washington University in St Louis St Louis, MO, United States	Amelioration of enteric nervous system defects via gene therapy in CLN1 disease mice
	Hemanth Nelvagal Washington University in St. Louis St. Louis, MO, United States	Efficacy of recombinant human PPT1 enzyme replacement therapy in mouse and sheep models of CLN1 disease
	Miriam Nickel University Medical Center Hamburg-Eppendorf Hamburg, Germany	Hamburg iNCL scale: A new tool for the quantitative description of disease progression in infantile CLN1 patients
	Angela Schulz University Medical Center Hamburg-Eppendorf Hamburg, Germany	Natural history of CLN7 disease: Quantitative prospective assessment of disease characteristics and rate of progression
	Live Moderated Q&A	*Jonathan Cooper, Hemanth Nelvagal, Miriam Nickel, and Angela Schulz*
11:30	Break, Exhibits and Satellite Symposia	
1:00	Troy Lund University of Minnesota Minneapolis, MN, United States	Bone marrow and umbilical cord blood are equivalent stem cell sources for Hurler syndrome
	Igor Nestrasil University of Minnesota Minneapolis, MN, United States	Quantitative brain MRI morphology in severe and attenuated forms of mucopolysaccharidosis type I
	Lynda Polgreen The Lundquist Institute at Harbor-UCLA Torrance, CA, United States	Anthropometric and joint deficits in children with mucopolysaccharidosis despite current treatments: A 10-year multi-site longitudinal study
	Tong Zhang Seattle Children's Research Institute Seattle, WA, United States	Development of multiplexed proteomic quantification of GAA and IDUA signature peptides in dried blood spots and buccal swabs by immuno-SRM-MS/MS for second-tier screening of Pompe disease and Hurler syndrome
	Live Moderated Q&A	*Troy Lund, Igor Nestrasil, Lynda Polgreen, and Tong Zhang*
2:00	Erin Huggins Duke University Durham, NC, United States	Early clinical phenotype of late-onset Pompe disease: Lessons learned from newborn screening
	Lisa Berry Cincinnati Children's Hospital Medical Center Cincinnati, OH, United States	Newborn screening for lysosomal disorders: The Ohio experience
	Haiyan Fu University of North Carolina at Chapel Hill Chapel Hill, NC, United States	Transient depletion of pre-existing antibodies for efficient AAV gene delivery
	Jillian Gallagher University of Massachusetts Medical School Worcester, MA, United States	Sialidosis: From gene editing to gene therapy **2022 Young Investigator Award Recipient*
	Live Moderated Q&A	*Erin Huggins, Lisa Berry, Haiyan Fu, and Jillian Gallagher*
3:00	Poster Session in the Exhibit Hall	
5:30	Satellite Symposia	

WORLD*Symposium*™ 2022 Preliminary Program*
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Wednesday, February 9, 2022
Clinical Applications

7:30	Chester B. Whitley University of Minnesota Minneapolis, MN, United States	Welcome and Keynote Speaker Introduction
7:35	Tippi MacKenzie University of California, San Francisco San Francisco, CA, United States	Prenatal enzyme replacement therapy for lysosomal disorders: Launching a phase I clinical trial
8:00	Simon Jones St. Mary's Hospital Manchester, United Kingdom	Clinical trial update: *Ex-vivo* autologous haematopoietic stem cell gene therapy in MPS IIIA
	Paul Harmatz UCSF Benioff Children's Hosptial Oakland, CA, United States	RGX-121 gene therapy for the treatment of severe mucopolysaccharidosis type II (MPS II): Interim analysis of data from the first in-human study
	Maurizio Scarpa Regional Coordinator Centre for Rare Diseases, University Hospital of Udine Udine, Italy	Continued improvement in pulmonary outcomes in 3 clinical trials of olipudase alfa in children and adults with chronic acid sphingomyelinase deficiency treated for 2 to 6.5 years
	Raymond Wang CHOC Children's Hospital Orange, CA, United States	RGX-111 gene therapy for the treatment of severe mucopolysaccharidosis type I (MPS I): Interim analysis of data from the first in-human study
	Live Moderated Q&A	*Simon Jones, Paul Harmatz, Maurizio Scarpa, and Raymond Wang*
9:00	Roberto Giugliani Federal University of Rio Grande do Sul Porto Alegre, Brazil	Long term efficacy and safety of pabinafusp-alfa (JR-141) in Hunter syndrome (MPS-II): 104-week data from the clinical trials in Japan and Brazil
	Robin Lachmann Charles Dent Metabolic Unit, National Hospital for Neurology and Neurosurgery London, United Kingdom	Sustained and continued improvements in pulmonary function, hepatosplenomegaly, dyslipidemia, and disease biomarkers in 5 adults with chronic acid sphingomyelinase deficiency after 6.5 years of olipudase alfa enzyme replacement therapy
	Ian O'Connor Medical University of South Carolina College of Medicine Charleston, SC, United States	Incidental diagnosis of lysosomal diseases by expanded carrier screening and direct-to-consumer genetic testing
	Linda Scheffers Erasmus MC Rotterdam, Netherlands	Effects of enzyme replacement therapy on cardiac function and structure in classic infantile Pompe disease: Up to 22 years of follow-up **2022 Young Investigator Award Recipient*
	Live Moderated Q&A	*Roberto Giugliani, Robin Lachmann, Ian O'Connor, and Linda Scheffers*
10:00	Break & Exhibits	
10:30	Yin-Hsiu Chien National Taiwan University Hospital Taipei, Taiwan	Immunogenicity of cipaglucosidase alfa/miglustat versus alglucosidase alfa/placebo in late-onset Pompe disease (LOPD): A phase III, randomized study (PROPEL)
	Eric Mallack Weill Cornell Medicine New York, NY, United States	A phase 1/2 open-label, multicenter, dose ranging and confirmatory study to assess the safety, tolerability and efficacy of PBKR03 administered to pediatric subjects with early infantile Krabbe disease (globoid cell leukodystrophy; GALax-C)

Wednesday, February 9, 2022
Clinical Application (*cont.*)

	Shaun Brothers University of Miami Miller School of Medicine Miami, FL, United States	Development of formulated resveratrol (micellar resveratrol) as a small molecule treatment for MPS I
	Jerry Vockley University of Pittsburgh Pittsburgh, PA, United States	An open-label, phase 1/2 trial of gene therapy 4D-310 in adult males with Fabry disease
	Live Moderated Q&A	*Yin-Hsiu Chien, Eric Mallack, Shaun Brothers, and Jerry Vockley*
11:30	Break, Exhibits and Satellite Symposia	
1:00	Cynthia Tifft National Human Genome Research Institute Bethesda, MD, United States	Phase 1/2 trial of AXO-AAV-GM1 (AAV9-GLB1) gene therapy for infantile- and juvenile-onset GM1 gangliosidosis
	Jeanine Jarnes University of Minnesota Minneapolis, MN, United States	Phase 1/2 open-label, multi-center study to assess the safety, tolerability and efficacy of a single dose of PBGM01 delivered into the cisterna magna of subjects with type 1 (early onset) and type 2a (late onset) infantile GM1 gangliosidosis
	Saima Kayani University of Texas Southwestern Medical Center Dallas, TX, United States	Preliminary safety data of a phase I first in-human clinical trial support the use of high dose intrathecal AAV9/CLN7 for the treatment of patients with CLN7 disease
	Stephanie Cherqui University of California, San Diego La Jolla, CA, United States	Hematopoietic stem cell gene therapy for cystinosis: Updated results from a phase I/II clinical trial
	Live Moderated Q&A	*Cynthia Tifft, Jeanine Jarnes, Saima Kayani, and Stephanie Cherqui*
2:00	Ecenur Tuc Bengur University of Pittsburgh Medical Center – Children's Hospital of Pittsburgh Pittsburgh, PA, United States	Psychosine predicts age of onset in babies with Krabbe disease
	Alexander Broomfield Royal Manchester Children's Hospital Manchester, United Kingdom	Neurocognitive outcome in mucopolysaccharidosis type I (Hurler phenotype) post HSCT
	Shoshana Revel-Vilk Shaare Zedek Medical Center Jerusalem, Israel	Markers of inflammation and alpha degranulation defect of platelets in patients with Gaucher disease
	Michal Becker-Cohen Shaare Zedek Medical Center Jerusalem, Israel	An 18-month report on the safety and efficacy of rapid intravenous velaglucerase alfa infusions in naïve patients with Gaucher disease
	Live Moderated Q&A	Ecenur Tuc Bengur, Alexander Broomfield, Shoshana Revel-Vilk, and Michal Becker-Cohen
3:00	Poster Session in the Exhibit Hall	
5:30	Satellite Symposia	

Thursday, February 10, 2022
Contemporary Forum *(not available for Continuing Education Credits)*

7:30	Chester B. Whitley University of Minnesota Minneapolis, MN, United States	Welcome and New Treatment Awards
8:00	Anna Bakardjiev Denali Therapeutics South San Francisco, CA, United States	Interim 49-week results of a phase 1/2 study of intravenous DNL310 (brain-penetrant enzyme replacement therapy) in MPS II
	Jacinthe Gingras Homology Medicines Bedford, MA, United States	Clinical trial design for HMI-203 investigational gene therapy for mucopolysaccharidosis type II (MPS II) informed by cross-correction potential and KOL input
	Nidal Boulos REGENXBIO Rockville, MD, United States	Identification of a biomarker that differentiates neuronopathic forms of MPS I and MPS II
	Laura Smith Homology Medicines Bedford, MA, United States	Summary of nonclinical data for gene therapy developmental candidate HMI-203 for mucopolysaccharidosis type II (MPS II, or Hunter syndrome)
	Live Moderated Q&A	*Anna Bakardjiev, Jacinthe Gingras, Nidal Boulos, and Laura Smith*
9:00	Rebeca Choy Maze Therapeutics, Inc South San Francisco, CA, United States	In-vitro characterization of MZE001, an orally active GYS1 inhibitor to treat Pompe disease
	Maria Praggastis Regeneron Pharmaceuticals Tarrytown, NY, United States	BBB-targeted GAA delivered as gene therapy treats CNS and muscle in Pompe disease model mice
	Julie Ullman Maze Therapeutics South San Francisco, CA, United States	Substrate reduction therapy for Pompe disease: Small molecule inhibition of glycogen synthase 1 in preclinical models
	Niek van Til AVROBIO, Inc. Cambridge, MA, United States	Long-term hematopoietic stem cell lentiviral gene therapy rescues neuromuscular manifestations in preclinical study of Pompe disease mice
	Live Moderated Q&A	*Rebeca Choy, Maria Praggastis, Julie Ullman, and Niek van Til*
10:00	Break & Exhibits	
10:30	Maria Escolar University of Pittsburgh Pittsburgh, PA, United States	FBX-101, an intravenous AAV gene replacement therapy given after infusion of hematopoietic stem cells, extends efficacious dose ranging and corrects disease manifestations in Krabbe disease
	Russell Gotschall M6P Therapeutics St. Louis, MO, United States	M011: A novel highly phosphorylated β-glucocerebrosidase enzyme with broader tissue biodistribution for the treatment of Gaucher disease
	Erika Pearson Sigilon Therapeutics Cambridge, MA, United States	Development of a novel encapsulated non-viral cell-based, BBB-penetrant therapy for MPS I

Thursday, February 10, 2022
Contemporary Forum (*cont.*) *(not available for Continuing Education Credits)*

	Francois-Xavier Frapaise Lysogene Neuilly-sur-Seine, France	A study of intracisternal administration of adeno-associated viral vector serotype rh.10 carrying the human β-galactosidase cDNA for the treatment of GM1 gangliosidosis: Preliminary results of the safety cohort
	Live Moderated Q&A	*Maria Escolar, Russell Gotschall, Erika Pearson, and Francois-Xavier Frapaise*
11:30	Break, Exhibits and Satellite Symposia	
1:00	Christiane Hampe Immusoft Seattle, WA, United States	Iduronidase-transposed human B lymphocytes correct enzyme deficiency and glycosaminoglycan storage disease in immunodeficient mucopolysaccharidosis type I mice
	Andrew Hedman M6P Therapeutics St. Louis, MO, United States	A novel S1S3 phosphotransferase co-expression gene therapy platform for lysosomal disorders
	Elizabeth Hwang-Wong Regeneron Pharmaceuticals Tarrytown, NY, United States	Defining phenotype reversibility in lysosomal disease: Leveraging a COIN model in mucopolysaccharidosis type VI (MPS VI)
	Leslie Jacobsen Neurogene Inc. New York, NY, United States	Efficacy of gene therapy in a CLN5 sheep model using a dual route of administration supports a first-in-human clinical trial
	Live Moderated Q&A	*Christiane Hampe, Andrew Hedman, Elizabeth Hwang-Wong, and Leslie Jacobsen*
2:00	Kyle Landskroner Azafaros Basel, Switzerland	Characterization of AZ-3102, a novel brain-penetrant small molecule, in the Niemann-Pick disease type C mouse model
	Ralph Laufer Lysogene Neuilly-sur-Seine, France	AAVance gene therapy study in children with mucopolysaccharidosis type IIIA
	Mariana Loperfido AVROBIO Inc Cambridge, MA, United States	High-resolution cellular and molecular follow up of lysosomal disease patients treated with hematopoietic stem cell lentiviral gene therapy
	Luca Biasco AVROBIO, Inc. Cambridge, MA, United States	High throughput monitoring of safety, potency and stability of gene therapy cell products in lysosomal disease patients
	Live Moderated Q&A	*Kyle Landskroner, Ralph Laufer, Mariana Loperfido, and Luca Biasco*
3:00	Poster Session	
5:30	Satellite Symposia	

Friday, February 11, 2022
Late-Breaking Science *(not available for CE accreditation; CE credits for GCs may apply)*

6:45	Satellite Symposia	
8:00	Francesca Tucci IRCCS San Raffaele Scientific Institute Milan, Italy	First-in-human phase I/II clinical trial of hematopoietic stem and progenitor cell gene therapy for Hurler syndrome: Favorable safety profile and extensive metabolic correction
	Priya Kishnani Division of Medical Genetics, Duke University Medical Center Durham, NC, United States	Avalglucosidase alfa improves health-related quality of life (HRQoL) in patients with late-onset Pompe disease (LOPD) vs. alglucosidase alfa: Patient-reported outcome measures (PROMs) from the phase 3 COMET trial
	Derralynn Hughes Royal Free London NHS Foundation Trust London, United Kingdom	Safety and efficacy of FLT190 for the treatment of patients with Fabry disease: Results from the MARVEL-1 phase 1/2 clinical trial
	David Weinstein Passage Bio Philadelphia, PA, United States	Safety, biomarker and preliminary efficacy results following ICM administration of PBGM01 in children with late onset infantile GM1-gangliosidosis
	Live Moderated Q&A	*Priya Kishnani, Derralynn Hughes, and David Weinstein*
9:00	Taylor Fields IntraBio Ltd Oxford, United Kingdom	N-acetyl-l-leucine improves symptoms and functioning in Niemann-Pick disease type C (NPC) and GM2 gangliosidosis (Tay-Sachs disease & Sandhoff disease): Results from two parallel, multi-national, rater-blinded clinical trials
	Jaya Ganesh The Icahn School of Medicine at Mount Sinai New York, NY, United States	Preliminary results of the STAAR study, a phase I/II study of isaralgagene civaparvovec (ST-920) gene therapy in adults with Fabry disease
	Marie-Anne Colle Oniris, Vet School of Nantes Nantes, France	FOXO3a over-expression in Pompe disease alleviates muscle impairments autophagic buildup
	Live Moderated Q&A	*Taylor Fields, Jaya Ganesh, and Marie-Anne Colle*
10:00	Break	
10:15	Victoria Jensen Lacerta Therapeutics Alachua, FL, United States	Long-term correction of mucopolysaccharidosis type IIIB disease phenotype following central nervous system administration of AAV-NAGLU
	Naresh Kumar Meena National Institutes of Health Bethesda, MD, United States	Liver-directed and systemic AAV gene transfer approaches for Pompe disease therapy
	Kwi Hye Kim REGENXBIO Inc Rockville, MD, United States	Establishment of in vitro model of CLN2 retinopathy using human induced pluripotent stem cells

	John Mitchell Montreal Children's Hospital Montreal, QC, Canada	Long-term outcomes of MPS IVA patients treated with elosulfase alfa: Findings from the Morquio A Registry Study (MARS) after 6 years
	Live Moderated Q&A	*Victoria Jensen, Naresh Kumar Meena, Kwi Hye Kim, and John Mitchell*
11:15	Behzad Najafian University of Washington Seattle, WA, United States	Venglustat reduces globotriaosylceramide inclusions in skin arterial smooth muscle cells in treatment naive males with classic Fabry disease
	Jagdeep Walia Kingston Health Sciences Centre and Queen's University Kingston Kingston, ON, Canada	AZ-3102, a novel brain-penetrant small molecule, significantly improves survival of Sandhoff disease mice
	Markus Schwarz ARCHIMEDlife Vienna, Austria	High-risk population screening by differential diagnosis for mucopolysaccharidoses (MPSs)
	Live Moderated Q&A	*Behzad Najafian, Jagdeep Walia, and Markus Schwarz*
12:00	Meeting Adjourns	
12:00	Satellite Symposium	On Demand only

**The Preliminary Program is subject to change without notice. Any updates to the program will be posted on the website: worldsymposia.org*