UDNI
Undiagnosed
Diseases Network
INTERNATIONAL

12TH INTERNATIONAL CONFERENCE ON RARE AND UNDIAGNOSED DISEASES

THE PROGRAM

UDNI 2023

OCTOBER 22-23, 2023
TBILISI, GEORGIA

www.udninternational.org

CONFERENCE CHAIRS

Oleg Kvlividze
Georgian Foundation for Genetic and Rare Diseases (GeRaD), Georgia

Tinatin Tkemaladze
Tbilisi State Medical University (TSMU), Georgia

Willian A. Gahl
National Institute of Health (NIH), USA

INTERNATIONAL SCIENTIFIC COMMITTEE

Willian A. Gahl
National Institute of Health (NIH), USA

LOCAL SCIENTIFIC COMMITTEE

Eka Kvaratskhelia
Tbilisi State Medical University (TSMU), Georgia

PROGRAM FOR SATURDAY OCTOBER 21, 2023

11.00-14.00	ST-01	TOWARDS INTERNATIONAL INTEGRATION	LIVING ROOM
15.00-18.00	ST-02	RARE AND UNDIAGNOSED DISEASES: BUILDING NEIGHBOR BRIDGES (ACIBADEM)	LIVING ROOM

PROGRAM FOR SUNDAY OCTOBER 22, 2023

10.00-12.20	SU-01	DIAGNOSIS OF RARE AND UNDIAGNOSED DISEASES - COMMON CHALLENGES	ADJARA HALL
12.30-13.30	LB-01	LUNCH BREAK	LAUNGE AREA
12.30-13.30	JN-01	RARE - NEW JOURNAL PRESENTATION	GLASS ROOM
13.30-14.30	ST-03	EVOLVING TREATMENT LANDSCAPE IN SMA (NOVARTIS)	LIVING ROOM
13.30-15.50	SU-02	NOVEL DIAGNOSTICS AND THERAPEUTICS FOR RARE DISEASES	ADJARA HALL
16.00-16.30	CB-01	COFFEE BREAK	LAUNGE AREA
16.30-17.30	ST-04	BRINGING CLINICAL VALUE THROUGH VALIDATED ANALYSIS (BLUEPRINT GENETICS)	LIVING ROOM
16.30-18.30	SU-03	RARE AND UNDIAGNOSED DISEASES WORLDWIDE	ADJARA HALL
18.40-19.30	PS-01	POSTER SESSION	GREEN ROOM
20.30-23.00	OC-01	OPENING CEREMONY, WELCOME RECEPTION	CITY COUNCIL

PROGRAM FOR MONDAY OCTOBER 23, 2023

09.00-09.55	MO-01	NOVEL DIAGNOSTICS AND THERAPEUTICS FOR RARE DISEASES	ADJARA HALL
10.00-10.54	MO-02	NEW DISEASES; SOLVED AND UNSOLVED CASES	ADJARA HALL
11.00-12.20	MO-03	INTERNATIONAL INITIATIVES ON RARE AND UNDIAGNOSED DISEASES	ADJARA HALL
12.20-13.20	LB-02	LUNCH BREAK	LAUNGE AREA
13.20-15.00	ST-05	WHAT RARE DISEASES ARE HIDING BEHIND CHOLESTASIS IN CHILDREN? (BIOMEDICA)	TERRACE ROOM
13.20-16.15	RP-01	UDNI COMMITTEE & WORKING GROUP REPORT	ADJARA HALL
16.30-17.00	CB-02	COFFEE BREAK	LAUNGE AREA
17.00-19.15	GE-01	GEORGIAN SESSION	ADJARA HALL
17.00-18.00	BM-01	UDNI BUSINESS MEETING	LIVING ROOM
20.00-23.30	GD-01	GALA DINNER	

SUTERDAY
OCT. 21 2023

PRE-CONGRESS DAY

ST-01 TOWARDS INTERNATIONAL INTEGRATION (PATIENTS MEETING SUPPORTED BY WILHELM FOUNDATION, EURORDIS AND NORD)

11.00-14.00

ST-02 RARE AND UNDIAGNOSED DISEASES: BUILDING NEIGHBOR BRIDGES / TR-AZ-GEO (SUPPORTED BY ACIBADEM)

15.00-18.00

SUNDAY
OCT. 22 2023

SCIENTIFIC PROGRAM, DAY 1

08.30 REGISTRATION

09.30-10.00 CONFRENCE OPENING & WELCOME

SU-01 DIAGNOSIS OF RARE AND UNDIAGNOSED DISEASES - COMMON CHALLENGES
Moderators: Siddharth Banka (UK), Tinatin Tkemaladze (Georgia)

10.00-10.20
Sofia Douzgou Houge
Haukeland University Hospital, Norway
THE POST-EXOME CLINIC: IMPROVING THE IMPACT OF EXOME SEQUENCING FOR DEVELOPMENTAL DISORDERS

10.20-10.40
Lama Al-Abdi
King Faisal Specialist Hospital & Research Centre, Saudi Arabia
THE DIAGNOSTIC IMPLICATIONS OF PITFALLS IN CAUSAL VARIANT IDENTIFICATION BASED ON 4,577 MOLECULARLY CHARACTERIZED FAMILIES

10.40-11.00
Steven Laurie
The Barcelona Institute of Science and Technology, Spain
HOW CAN WE REACH A DIAGNOSIS FOR MORE INDIVIDUALS WITH A RARE CONDITION, THE SOLVE-RD PERSPECTIVE

11.00-11.20
Wendy van Zelst-Stams
Radboudumc, The Netherlands
DNA FIRST STRATEGIES TO REDUCE THE DIAGNOSTIC ODYSSEY IN RARE DISEASE PATIENTS

11.20-11.40
Gunnar Douzgos Houge
Haukeland University Hospital, Bergen, Norway
INTERPRETATION AND CLASSIFICATION OF GENETIC VARIANTS

11.40-12.00
Michael Brudno
University of Toronto, Canada
CAPTURING AND SHARING DATA ON RARE DISEASES PATIENTS IN THE CANADIAN CARE4RARE PROJECT

12.00-12.20	**Yasemin Alanay** Acibadem University, Turkey RARE DISEASES IN DIVERSE POPULATIONS: CLINICAL WGS EXPERIENCE FROM ISTANBUL, AN EMA HUB
LB-01	**LUNCH BREAK**
12.30-13.30	
JN-01	**RARE: OPEN RESEARCH IN RARE DISEASES - NEW JOURNAL PRESENTATION**
12.30-13.30	
ST-03	**EVOLVING TREATMENT LANDSCAPE IN SMA (SATELLITE SPONSORED BY NOVARTIS)**
13.30-14.30	
SU-02	**NOVEL DIAGNOSTICS AND THERAPEUTICS FOR RARE DISEASES** **Moderators: Domenica Taruscio (Italy), Eka Kvaratskhelia (Georgia)**
13.30-13.50	**Ari Zimran** Gaucher Clinic at Shaare Zedek Medical Center, Israel THERAPY IN GBA1 RELATED PARKINSON: THE NEVER-ENDING STORY
13.50-14.10	**Manuel Schiff** Hôpital Necker-Enfants Malades, France INHERITED (LIVER) METABOLIC DISEASES: IS THERE A THERAPEUTIC UNMET NEED? EXAMPLES OF PRECLINICAL DATA FOR GENE THERAPY IN MAPLE SYRUP URINE DISEASE (MSUD)
14.10-14.30	**Siddharth Banka** University of Manchester, UK DISSECTING CLINICAL, GENETIC, AND MECHANISTIC HETEROGENEITY OF NON-MUSCLE ACTINOPATHIES
14.30-14.50	**Arndt Rolfs** University of Rostock, Germany IMPORTANCE OF WHOLE GENOME SEQUENCING FOR THE EARLY IDENTIFICATION OF RARE GENETIC DISORDERS
14.50-15.10	**Yair Anikster** Sheba-Tel HaShomer Hospital, Israel NOVEL MITOCHONDRIAL AUGMENTATION THERAPY - FROM BENCH TO BEDSIDE
15.10-15.30	**Stephen Meyn** University of Wisconsin-Medison, USA BEYOND EXOMES: NEW OPPORTUNITIES FOR UNDIAGNOSED DISEASE PROGRAMS
15.30-15.50	**Alain Hovnanian** Hôpital Necker-Enfants Malades, France CURRENT AND EMERGING TREATMENTS FOR INHERITED EPIDERMOLYSIS BULLOSA

CB-01 COFFEE BREAK

16.00-16.30

ST-04 BRINGING CLINICAL VALUE THROUGH VALIDATED ANALYSIS (SATELLITE SPONSORED BY BLUEPRINT GENETICS)

16.30-17.30

SU-03 RARE AND UNDIAGNOSED DISEASES WORLDWIDE

Moderators: William Gahl (USA), Oleg Kvlividze (Georgia)

16.30-16.50 **Milan Macek**
Charles University Hospital, Czech Republic

UNDIAGNOSED RARE DISEASES IN THE CZECH REPUBLIC: ACTIVITIES ON THE NATIONAL COORDINATION CENTRE FOR RARE DISEASES AND MULTIDISCIPLINARY NATIONWIDE COLLABORATION

16.50-17.10 **Tinatin Tkemaladze**
Tbilisi State Medical University, Georgia

Oleg Kvlividze
GeRaD, Georgia

RARE DISEASES IN GEORGIA: ACHIEVEMENTS AND CHALLENGES

17.10-17.30 **Salman Kirmani**
Aga Khan University, Pakistan

DIAGNOSING AND MANAGING RARE DISEASES IN LIMITED RESOURCE SETTINGS: PAKISTANI EXPERIENCE

17.30-17.50 **Natalia Samonenko**
Center of Orphan Disease and Gene Therapy; I NSCH “OKHMATDYT”, Ukraine

DIAGNOSTICS AND TREATMENT OF ORHAN DESEASES IN UKRAINE

17.50-18.10 **Ratna Puri**
Institute of Medical Genetics & Genomics, India

THE LANDSCAPE WITHIN THE INDIAN UNDIAGNOSED DISEASES PROGRAM: INSIGHTS & CHALLENGES

18.10-18.30 **Ugur Özbek**
Acibadem University, Turkey

RARE AND UNDIAGNOSED DISEASES: TURKISH PERSPECTIVES

PS-01 POSTER SESSION

18.40-19.30

OC-01 OPENING CEREMONY, WELCOME RECEPTION

20.30-23.00

MONDAY
OCT. 23
2023

SCIENTIFIC PROGRAM, DAY 2

M0-01 NOVEL DIAGNOSTICS AND THERAPEUTICS FOR RARE DISEASES

Moderator: David Adams (USA)

09.00-09.15
Vorasuk Shotelersuk
Chulalongkorn University, Thailand
APPLICATIONS OF LONG-READ SEQUENCING FOR RARE DISEASES

09.15-09.25
Tahsin Stefan Barakat
Erasmus MC University Medical Center, The Netherlands
SOLVING UNDIAGNOSED DISEASE BEYOND THE EXOME: FUNCTIONAL ENHANCERS WITH MEDICAL RELEVANCE IDENTIFIED BY COMPUTATIONAL ANALYSIS AND CHIP-STARR-SEQ IN NEURAL CELL MODELS ENABLE PRIORITIZING NON-CODING VARIANTS FROM PATIENT WHOLE GENOME SEQUENCING STUDIES

09.25-09.35
Shinya Yamamoto
Baylor College of Medicine, USA
FUNCTIONAL STUDIES USING DROSOPHILA SUPPORT CLINICAL DIAGNOSIS AND PHENOTYPIC EXPANSION: BMPR2 IN NEURODEVELOPMENTAL DISORDERS

09.35-09.45
Sandro Surmava
Tbilisi State Medical University, Georgia
EPIGENETICS IN RARE DISEASES: THE ROLE IN CYSTIC FIBROSIS

09.45-09.55
Rachel Rock
Metabolic Diseases Clinic, Edmond and Lily Safra Children's Hospital, Sheba Medical Center, Israel
PHENYLALANINE TREATMENT IN 8-MONTH OLD GIRL WITH MITOCHONDRIAL FARS2 DEFICIENCY

M0-02 NEW DISEASES; SOLVED AND UNSOLVED CASES

Moderator: May Christine Malicdan (USA)

10.00-10.08
Vorasuk Shotelersuk
Chulalongkorn University, Thailand
NEW DISEASE DISCOVERED VIA UDNI COLLABORATION: BENIGN ADULT FAMILIAL MYOCLONIC EPILEPSY TYPE 8 (BAFME8)

10.08-10.16
Ben Pode-Shakked
Edmond and Lily Safra Children's Hospital, Sheba Medical Center, Israel
PERCC1-ASSOCIATED CONGENITAL ENTEROPATHY: DELINEATING THE NATURAL HISTORY OF A NEW DISORDER OF ENTEROENDOCRINE CELL FUNCTION

10.16-10.24
Guida Landoure
Faculté de Médecine et d'Odontostomatologie, USTTB, Mali
UNRAVELING THE GENETIC BASIS OF RARE HEREDITARY NEUROLOGICAL DISEASES IN MALI

10.24-10.30
Anastasia Sukhiashvili
Tbilisi State Medical University, Georgia
THE CASE OF RUBINSTEIN-TAYBI SYNDROME - HUNTING FOR HIDDEN MUTATION

10.30-10.38

Yehoshua (Joshua) Manor
Edmond and Lily Safra Children's Hospital, Sheba Medical Center, Israel

FAILURE TO THRIVE, ICHTHYOSIS, DEAFNESS, AND ENDOCRINOPATHIES IN AN INFANT WITH A NOVEL BIALLELIC AP1B1 MUTATION CAUSING ABNORMAL INTRACELLULAR ATP7A TRAFFICKING

10.38-10.46

Shmuel Prints
Clalit Health Services, Israel

THE UNDIAGNOSED DISEASE MASQUERADE: A FAMILY CASE OF SEVERE ASTHMA SUCCESSFULLY TREATED WITH MEPOLIZUMAB

10.46-10.54

Roberto Giugliani
Federal University, Rio Grande do Sul (UFRGS), Brazil

UNDIAGNOSED SYNDROMIC HYPERINSULINEMIC HYPOGLYCEMIA IN AN INFANT

MO-03 INTERNATIONAL INITIATIVES ON RARE AND UNDIAGNOSED DISEASES

Moderators: Vorasuk Shotelersuk (Thailand)

11.00-11.15

David Pearce
Sanford School of Medicine, University of South Dakota, USA

THE INTERNATIONAL RARE DISEASE RESEARCH CONSORTIUM (IRDIRC): MAKING RARE DISEASE RESEARCH EFFORTS MORE EFFICIENT AND COLLABORATIVE, AROUND THE WORLD

11.15-11.30

Yanis Mimouni
EJP RD, France

CONSOLIDATING THE RARE DISEASES RESEARCH ECOSYSTEM: A MULTISTAKEHOLDER AND MULTIDIRECTIONAL APPROACH

11.30-11.45

Tania Simoncelli
Chan-Zuckerberg Initiative, USA

BEYOND ENGAGEMENT: PATIENT COMMUNITIES AS CRITICAL DRIVERS OF RARE DISEASES RESEARCH?

11.45-12.00

Aung Min Saw
Syndrome Without a Name (SWAN), Wales, UK

Matthew Spencer
NHS Wales, Swansea University, UK

CHALLENGES UNMASKED: NAVIGATING THE COMPLEXITIES OF ADULT RARE AND UNDIAGNOSED DISEASE CARE

12.00-12.10

Jaeso Cho
Seoul National University Hospital, Republic of Korea

CLINICAL UTILITY OF GENETIC DIAGNOSIS IN ADULTS WITH UNDIAGNOSED DISEASE: AN EXPERIENCE FROM KOREAN ADULT UNDIAGNOSED DISEASE PROGRAM

12.10-12.20

Vincenzo Nigro
Telethon Institute of Genetics and Medicine, Italy

A SYSTEMATIC APPROACH FOR THOUSAND SEVERE UNSOLVED PEDIATRIC CONDITIONS: RESULTS FROM THE TELETHON UNDIAGNOSED DISEASE PROGRAM

LB-02 LUNCH BREAK

12.20-13.20

ST-05 WHAT RARE DISEASES ARE HIDING BEHIND CHOLESTASIS IN CHILDREN? (SATELLITE SPONSORED BY BIOMEDICA)

13.20-15.00

RP-01 UDNI COMMITTEE & WORKING GROUP REPORTS
Moderator: William Gahl (USA)

13.20-13.30	William Gahl (USA) REVIEW OF UDNI AND CURRENT LEADERSHIP
13.30-13.40	Eric Klee (USA) MEMBERSHIP; TRAINEE CATEGORY/UPDATE
13.40-13.50	Janine Lewis (USA), Stephanie Broley (Australia) GENETIC COUNSELING WORKING GROUP
13.50-14.00	Marco Salvatore (Italy), Gianluca Ferrari (Italy), Domenica Taruscio (Italy) COMMUNICATIONS/WEBSITE
14.00-14.15	Domenica Taruscio (Italy), Manuel Posada (Spain), Samuel Wiafe (Ghana), Olaf Bodamer (USA) DEVELOPING NATIONS WORKING GROUP
14.15-14.25	Bela Melegh (Hungary), Domenica Taruscio (Italy), Bruce Korf (USA) EDUCATION WORKING GROUP/UEMS/MEDICAL COMPETENCE AND MEDICAL SPECIALTY
14.25-14.35	Gulcin Gumus (Spain), Helene Cederroth (Sweden), Debbie Drell (USA) PATIENT ENGAGEMENT PLUS
14.35-14.45	Ann Nordgren (Sweden), Emma Palmer (Australia), Lorenzo Botto (USA) DIAGNOSTICS WORKING GROUP: LINKING TO HACKATHONS AND EMERGING TECHNOLOGY
14.45-15.15	Mikk and Helene Cederroth (Sweden), Ann Nordgren (Sweden), Salman Kirmani (Pakistan), Samuel Wiafe (Ghana), Aime Lumaka (DR Congo), Wendy van Zelst-Stams (The Netherlands) HACKATHON UPDATE AND FOLLOWUP; VIDEO SUMMARY OF STOCKHOLM HACKATHON IMPRESSIONS FROM CHAMPIONS Wendy van Zelst-Stams (The Netherlands) FUTURE HACKATHON (IN NIJMEGEN, THE NETHERLANDS)
15.25-15.45	David Adams (USA), Alistair Ward (USA) DATA SHARING COMMITTEE: NEW ANALYTICAL TOOLS, LONG READS, ETC. CALYPSO AND IOBIO TOOLS IN THE UDN
15.45-16.15	William Gahl (USA), Helene and Mikk Cederroth (Sweden), UDNI DNWG: Samuel Wiafe (Ghana), Salman Kirmani (Pakistan), Aime Lumaka (DR Congo), Guida Landoure (Mali) CHAMPIONS INITIATIVE: INTRODUCTION / GHANA, PAKISTAN, DR CONGO, MALI INTERCONTINENTAL EXCHANGE DISCUSSION

CB-02 COFFEE BREAK

16.30-17.00

GE-01 CURRENT ISSUES OF RARE AND UNDIAGNOSED DISEASES (GEO)

Moderators: Ivane Chkhaidze (Georgia), Maia Kherkheulidze (Georgia)

17.00-17.15
Nana Tatishvili
D. Tvildiani Medical University; M. Iashvili Children's Central Hospital, Georgia
SUCCESS AND CHALLENGES IN THE TREATMENT OF SMA: GEORGIAN EXPERIENCE

17.15-17.30
Mariam Ghughunishvili
G. Zhvania Pediatric Academic clinic, Georgia
HOPE ON THE HORIZON: ALAGILLE SYNDROME TREATMENT ADVANCEMENTS

17.30-17.45
Sophia Bakhtadze
Tbilisi State Medical University; G. Zhvania Pediatric Academic clinic, Georgia
NONEPILEPTIC PAROXISMS IN CHILDREN

17.45-18.00
Lali Margvelashvili
Center of Medical Genetics and Laboratory Diagnostics, Georgia
RARE DISEASES OUTPATIENT MONITORING PROGRAM IN GEORGIA - ACHIVEMENTS, CHALLENGES AND EXPECTATIONS

18.00-18.15
Teona Shatirishvili
D. Tvildiani Medical University; M. Iashvili Children's Central Hospital, Georgia
MODERN TREATMENT OF DUCHENNE MUSCULAR DYSTROPHY

18.15-18.30
Dodo Agladze
P. Shotadze Tbilisi Medical Academy; Center of Medical Genetics and Laboratory Diagnostics, Georgia
CF IN GEORGIA, CURRENT TREATMENT AND FUTURE PERSPECTIVE

18.30-18.45
Jenara Kristesashvilil
Faculty of Medicine, I. Javakhishvili State University; Reproductive Medical Center Universe, Georgia
RARE FORMS OF CONGENITAL SEX DEVELOPMENT DISORDERS (DSD)

18.45-19.00
Maia Kherkheulidze
Tbilisi State Medical University; G. Zhvania Pediatric Academic clinic, Georgia
GALACTOSEMIA: TREATMENT RECOMMENDATIONS

19.00-19.15
Ekaterine Kvatatskhelia
Ilia State University; Davit Metreveli Medical Center, Georgia
TREATMENT ASPECTS OF CONGENITAL ADRENAL HYPERPLASIA

BM-01 UDNI BUSINESS MEETING

Moderator: William Gahl (USA)

17.00-18.00

1. BOARD TO APPROVE NEW MEMBERS;
2. VOTES: 2024 MEETING; RARE AFFILIATION;
3. 2024: SEOUL, SEPT. 5-7;
4. 2025: CALL FOR PROPOSALS;
5. BOD TURNOVER DISCUSSION; CHARTER;
6. RARE JOURNAL AFFILIATION;
7. ISSUES ARISING;
8. PREVIOUS TOPICS FOR DISCUSSION.

GD-01 GALA DINNER (KINDLY SPONSORED BY THE WILHELM FOUNDATION)

20.00-23.30

MAIN ORGANIZERS

CO-ORGANIZERS

SPONSORS

PLATINUM

GOLD

SILVER

BRONZE

Supported by Shota Rustaveli National Science Foundation of Georgia [MG-ISE-23-1447]

13.5
CME
Credits