



## **Programme**

**Republican Conference with international participation «Rare Diseases: from awareness to action»**

**February 27-28**

**March 01, 2025**

**Venue: 53 Kabanbai Batyr Avenue, Nazarbayev University, Astana**

**Astana city**

<b>Time</b>	
7.00- 8.00	<b>Registration</b>
<b>Opening ceremony</b>	
8.00 – 8.10	Welcoming speech <b>Akmaral Alnazarova</b> Minister of Health Republic of Kazakhstan
8.10 – 8.20	Welcoming speech <b>Pya Yuriy</b> University Medical Center, CEO Professor, member of the National Academy of Sciences Republic of Kazakhstan.
<b>Plenary Session</b> <b>Main conference hall.</b> “Orphan Diseases: Unite forces.” <b>Moderators:</b> Ivanova-Razumova Tatyana, Tuleutaev Ernas	
8.20 – 8.35	National Center for Rare Diseases: Results, Prospects, and Plans. <b>Speaker: Ernas Tuleutaev</b> , Doctor of Medical Sciences (D.M.Sc.) Republican Center for Rare Diseases, CF UMC
8.35 – 8.50	Organization of Care for Patients with Rare Diseases in Kazakhstan. <b>Speaker: Tatyana Ivanova-Razumova</b> , PhD, MBA. Director of Pediatrics Department, "University Medical Center" Supported by AstraZeneca
08:50-09:20	Improving Outcomes in Duchenne Muscular Dystrophy Patients by Restoring Dystrophin Through Exon Skipping. <b>Speaker: Dr. Perry Shieh (UCLA, USA) – (Online)</b>
09:20-09:40	Challenges and Approaches in Transitioning Orphan Disease Patients to Adult Care. <b>Speaker: Zaure Dmitrievna Dushimova</b> , PhD, Deputy Head of Fundamental Medicine, Al-Farabi Kazakh National University – <i>(Online)</i> .
09:40-09:55	Organizing Care for Citizens with Rare Diseases in Kazakhstan. <b>Speaker: Bakhyt Madeniyetovna Baigalieva</b> , Founder of "Kobelek Balalar" Charity Foundation. Founder and Head of the Association of Legal Entities "Association for Assistance to Patients with Rare Diseases in the Republic of Kazakhstan" Member of the Board of Trustees of the Public Foundation "Qazaqstan Khalkyna" Member of the Local Bioethics Commission of the NAS "Astana International University"
09:55-10:10	Digital Monitoring Tools for Healthcare Entities in the Provision of Medical Care to Orphan Patients <b>Natalya Valeryevna Kil</b> Board Member of the DAMU Information Technology Center
10:10-10:30	Coffee break
10:30-11:10	Multidisciplinary approach in managing patients with rare diseases: Focus on MPS” at International Conference On Rare Diseases in Kazakhstan, taking place on Feb 27- Mar 1, 2025 <b>Prof. Florian Lagler</b> from Institute of Inherited Metabolic Diseases, Paracelsus Medical University, Salzburg, Austria <i>(online) (Supported by BioMarin)</i>
11:10-11:25	Cystic Fibrosis in Kazakhstan: New Opportunities <b>Irina Yuryevna Mukatova</b> Professor, Doctor of Medical Sciences, NAO "Astana Medical University" Professor of the Department of Internal Diseases with a Course in Nephrology,

	Hematology, Allergology, and Immunology Chair of the Association of Respiratory Medicine Specialists
11:25-11:40	Organization of Medical Care for Children with Congenital Adrenal Cortex Dysfunction <b>Gulmira Kasenovna Abduakhasova</b> Assistant Professor, Department of Pediatric Diseases with a Course in Allergology, Hematology, and Endocrinology, NAO "Astana Medical University" Head Consulting Pediatric Endocrinologist of the Ministry of Health of the Republic of Kazakhstan
11:40-12:00	Palliative Care in Children <b>Nurila Amangaliyevna Maltabarova</b> Head of the Department of Pediatric Anesthesiology, Intensive Therapy, and SNP, NAO "Astana Medical University" Head Specialist in Pediatric Palliative Care of the Ministry of Health of the Republic of Kazakhstan
12:00-12:20	Modern Opportunities of ART (Assisted Reproductive Technology) in the Prevention of Monogenic Diseases and Chromosomal Pathology <b>Vyacheslav Notanovich Lokshin</b> Professor, Academician of the National Academy of Sciences of the Republic of Kazakhstan under the President of the Republic of Kazakhstan President of the Kazakhstan Association of Reproductive Medicine (KARM) General Director of the International Clinical Center for Reproductology "PERSONA" Head Reproductologist of the Ministry of Health of the Republic of Kazakhstan
12:20-13:00	<b>Moderator:</b> Tatyana Vladimirovna Ivanova-Razumova <b>Debate:</b> Home Respiratory Support – For or Against? <b>Participants:</b> N.A. Maltabarova, L.A. Tekebaeva
13:00-13:45	<b>Lunch</b>
	February 27, 2025 - Day 1 Main Conference Hall Neurology in the shadow: solutions for patients with rare diseases.
	<b>Moderators:</b> Jaksybaeva Altynshash, Lepesova Marjan, Myrzalievna Bahytgul
13.45 – 14.15	Consensus and Clinical Guidelines for Managing Patients with Spinal Muscular Atrophy: Criteria for Prescription, Monitoring, and Treatment Effectiveness Management of Neuromuscular Diseases: Experience in Kazakhstan. <b>Speaker: JJ. Andoni Urtizberea (France)</b> MD, MSc, Organizer of International Educational Programs in Myology, Head of the AcadeMyo - Summer School of Myology Project, Institute of Myology, Pitié-Salpêtrière University Hospital (Online)
14.15 – 14.30	Features of Implementing Gene Therapy Approaches in Medical Practice in Kazakhstan <b>Latina Aizhanovna Tekebaeva</b> Candidate of Medical Sciences, Head of the Pediatric Neurology Program, Corporate Fund "University Medical Center," National Research Center for Maternal and Child Health Head Pediatric Neurologist of the Ministry of Health of the Republic of Kazakhstan (Supported by Novartis)
14.30 – 14.50	Orphan Neuromuscular Diseases: Results of Genomic Studies on Kazakhstani Patients <b>Gulnara Salavatovna Svyatova</b> Doctor of Medical Sciences, Professor, LLP "Center for Molecular Medicine" President of the Public Association "Association of Medical Geneticists" (Almaty, Kazakhstan)
14.50 – 15.05	Neonatal Screening for Spinal Muscular Atrophy: Key Role in Determining Gene Therapy at the Preclinical Stage

	<p><b>Mirgul Fayzullinovna Bayanova</b> Head of the Clinical and Genetic Diagnostics Department, Corporate Fund "University Medical Center," Candidate of Medical Sciences (<i>Supported by Novartis</i>)</p>
15.05 – 15.20	<p>Patient Routing for Neuromuscular Diseases Using the Example of DMD in the Republic of Kazakhstan <b>Ernas Tleutaevich Tuleutaev</b> Head of the Republican Center for Rare Diseases, Corporate Fund "University Medical Center," Doctor of Medical Sciences (<i>Supported by PTC Therapeutics</i>)</p>
15.20 – 15.40	<p>Management of Duchenne Muscular Dystrophy: Therapy Approaches, Prescription Criteria, and Discontinuation of Pathogenetic Therapy <b>Altynshash Khairullayevna Jaksybayeva</b> President of the Public Association "Society of Pediatric Neurologists, Neurophysiologists, Psychiatrists, and Psychotherapists," Expert in Neuromuscular Diseases, Doctor of Medical Sciences (Almaty, Kazakhstan)</p>
15.40 – 16.00	<p>Application of Exon-Skipping Therapy in Patients with Duchenne Muscular Dystrophy in Kazakhstan: Experience of Children's City Hospital #2, Almaty Bakhytkul Jusupzhanovna Myrzaliyeva Master of Medical Sciences, Senior Lecturer at the Department of Neurology, KRMU, Neuromuscular Diseases Coordinator (Almaty, Kazakhstan)</p> <p><i>Co-Speaker: Olga Yakovlevna Zelman</i> Head of the Day Hospital, Children's City Hospital #2, Almaty</p>
16.00 – 16.20	<p><b>Coffee break</b></p>
	<p><b>Moderators:</b> Idrisova Zhanat, Dairbaeva Leila, Bembeeva Raisa</p>
16.20 – 16.40	<p>Myasthenia in Children: Management and Diagnostic Criteria Andoni Urtizberea (Paris, France) MD, MSc, Organizer of International Educational Programs in Myology, Head of the AcadeMyo - Summer School of Myology Project, Institute of Myology, Pitié-Salpêtrière University Hospital (<i>Online</i>)</p>
16.40 – 17.00	<p>Experience in Managing Patients with Neuromyelitis Optica Spectrum Disorder in Astana: Real Clinical Practice <b>Bagyzhan Rysbaevna Syzdykova</b> Head of the Neurology and Autoimmune Nervous System Diseases Center, "Multidisciplinary City Hospital No. 1," Astana Candidate of Medical Sciences, Head Neurologist of the Astana Health Department</p>
17.00-17.15	<p>Experience in Managing Patients with PN in NF1 at the Orphan Disease Center, UK Aksai, Almaty <b>Zhannat Rustemovna Idrisova (Almaty, Kazakhstan)</b> Doctor of Medical Sciences, Professor of the Department of Nervous Diseases, Kazakh National Medical University Pediatric Neurologist, Head of the Orphan Disease Center for Neurological Manifestations in Children (<i>Supported by AstraZeneca</i>)</p>
17.15 – 17.35	<p>Management of Multiple Sclerosis in Children <b>Raisa Tsendenkaevna Bembeeva (Moscow, Russia)</b> Doctor of Medical Sciences, Professor of the Department of Neurology, Neurosurgery, and Medical Genetics named after Academician L.O. Badalyan, Pirogov Russian National Research Medical University (Moscow, Russia)</p>
17.35-17.50	<p>Algorithms and Red Flags for Early Diagnosis of Epileptic Encephalopathies <b>Leila Oralgazievna Dairbayeva (Almaty/Astana, Kazakhstan)</b> MBA, Kazakhstan National League Against Epilepsy</p>

	Center for Neurology, Epileptology, and Rehabilitation Head Epileptologist of Almaty
17.50-18.05	Dravet Syndrome: Modern Approaches to Therapy and Diagnosis Based on Clinical Cases <b>Alisa Azamatovna Nauryzbayeva</b> Neurologist, Corporate Fund "University Medical Center"
18.05-18.20	Clinical Case of a Patient with AADC Deficiency in the Republic of Kazakhstan <b>Latina Aizhanovna Tekebaeva</b> Candidate of Medical Sciences, Head of the Pediatric Neurology Program, Corporate Fund "University Medical Center," National Research Center for Maternal and Child Health Head Pediatric Neurologist of the Ministry of Health of the Republic of Kazakhstan (Supported by PTC Therapeutics)
18:20-18:35	Rett Syndrome in the Structure of Rare Diseases <b>Anvar Taldybekovich Abzulin</b> Psychiatrist, Corporate Fund "University Medical Center"
18.35-18.50	Modern Approaches to the Diagnosis and Treatment of SMA in Children <b>Marzhan Makhmutovna Lepesova (Almaty, Kazakhstan)</b> Doctor of Medical Sciences, Professor, Head of the Department of Neurology, KazRosMedUniversity Chair of the Public Association "Association of Pediatric Neurologists" (Supported by Johnson & Johnson)
18:50-19:05	SMA Therapy – Multidisciplinary Approach and Adherence <b>Gulnar Amerzaevna Mukhambetova (Almaty, Kazakhstan)</b> Candidate of Medical Sciences, Professor of the Department of Nervous Diseases, Kazakh National Medical University named after S.D. Asfendiyarov (Supported by Johnson & Johnson)
19:05-19:20	Why It Is Important to Take Risdiplam Regularly: A Clinical Case Study <b>Guljaghan Saidullaevna Makhambetova (Shymkent, Kazakhstan)</b> Master of Medical Sciences, Head of the Neurology Department, City Children's Clinical Hospital, Shymkent
19:20-20:00	Discussion: Management of Rare Nervous System Diseases in Children. The Work of Multidisciplinary Teams from the Regions <b>Marzhan Makhmutovna Lepesova (Almaty, Kazakhstan)</b> Doctor of Medical Sciences, Professor, Head of the Department of Neurology, KRMU Chair of the "Association of Pediatric Neurologists"
	<b>February 27, 2025 - Day 1</b> <b>Conference hall № 1</b>
	<b>Orphan Diseases in adults: when rarity become reality</b>
	<b>Moderators: Rapiibekova Gulmira, Nabiev Altay, Durmanova Aigul</b>
13.45 – 14.05	Cushing's Disease <b>Zhanna Evgenievna Belaya (Moscow, Russia)</b> Doctor of Medical Sciences, Professor, Chief Researcher at the State Scientific Center of the Russian Federation, FSBI "National Medical Research Center of Endocrinology" of the Ministry of Health of the Russian Federation, Endocrinologist, Head of the Department of Osteoporosis and Osteopathy, Moscow (Supported by Recordati Rare Disease)
14.05 – 14.25	Unveiling the Heterogeneity of ALS Based on the Kazakhstan Registry Data 2023-2025 <b>Larisa Bolatovna Kuanova</b> Head of the Neurology Residency Program, Corporate Fund "University Medical Center" Professor, Doctor of Medical Sciences, Member of the American Academy of Neurology and the International ALS Association

14.25 – 14.45	Challenges in the Epidemiology, Diagnosis, and Treatment of Cushing's Disease and Syndrome in Kazakhstan <b>Anna Vikentyevna Bazarova</b> Candidate of Medical Sciences, Associate Professor of the Department of Internal Diseases with Courses in Gastroenterology, Endocrinology, and Pulmonology, NAO "Astana Medical University"
14.45 – 15.05	Pregnancy Planning for Monogenic Diseases Using Reproductive Technology Programs <b>Gulmira Kurbanovna Rapiibekova</b> Medical Director – Board Member, Corporate Fund "University Medical Center" Doctor of Medical Sciences, Obstetrician-Gynecologist
15.05 - 15.25	Challenges in the Diagnosis and Treatment of Rare Diseases with Microangiopathic Hemolysis During Pregnancy <b>Anton Anatolyevich Klodzinsky</b> Candidate of Medical Sciences, Medical Director of LLP "Hematology Center" Deputy Chairman of the Public Association "Kazakhstan Society of Hematologists"
15.25 – 15.45	Hereditary and Acquired Coagulopathies in Pregnant Women: How to Prevent Obstetric Hemorrhage <b>Makpal Sertaevna Abylkhalimova</b> Hematologist, LLP "Hematology Center"
15.45 – 16.00	Pulmonary Hypertension: Primary Pulmonary Hypertension During Pregnancy. <b>Speaker: Madina Sutralimova</b> , master of medical science, cardiologist, CF "UMC", Heart Center.
16.00 – 16.20	<b>Coffee break</b>
	<b>Moderators: Tuganbekova Saltanat, Gaipov Abdugappar</b>
16.20 – 16.40	Progress and Innovative Technologies in "Rare" and Hereditary Diseases in Nephrology <b>Saltanat Kenesovna Tuganbekova</b> Professor, Doctor of Medical Sciences, Highest Category Physician President of the National Kidney Foundation of the Republic of Kazakhstan Head Nephrologist of the Ministry of Health of the Republic of Kazakhstan
16.40 – 16.55	Genetic Landscape of ALS <b>Dieter Ritmacher</b> PhD, Vice Dean for Research and Postgraduate Programs, NU School of Medicine
16.55 – 17.10	Fabry Disease <b>Abduzhappar Erkinovich Gaipov</b> Professor, NU School of Medicine Nephrologist
17.10 – 17.25	Lipodystrophy: Continuity in Therapy <b>Aigul Kalibaevna Durmanova</b> Head of General Therapy Program 2, Corporate Fund "University Medical Center" Highest Category Physician, Doctor of Medical Sciences
17.25 – 17.40	Behchet's Disease. <b>Rysgul Malkenovna Aytzhanova</b> Head of General Therapy Program 1, Corporate Fund "University Medical Center" Highest Category Rheumatologist
17.40 – 18.00	Paroxysmal Nocturnal Hemoglobinuria (PNH) – Marchiafava-Micheli Disease <b>Zhanat Erkinovna Kuanshalieva</b> Highest Category Nephrologist, Corporate Fund "University Medical Center" Candidate of Medical Sciences

<b>February 27, 2025 - Day 1</b>	
<b>Conference hall № 2</b>	
<b>Orphan Immunological diseases</b>	
<b>Moderators: Kovzel Elena, Belevcev Mihail, Nima Rizai</b>	
13.45 – 14.10	<p>Congenital Immune System Defects: The Many Faces of Orphan Pathology. Standards and Protocols in Kazakhstan  <b>Elena Fedorovna Kovzel</b>  Professor, Corporate Fund "University Medical Center"  Head of the Clinical Immunology, Allergology, and Pulmonology Program  President of the Public Association "Kazakh Society of Immunodeficiencies – Association of Immunologists" Doctor of Medical Sciences, Head Allergist of the Republic of Kazakhstan</p>
14.10-14.30	<p>Congenital Immune System Defects: Advances in Diagnosis and Treatment  <b>Mikhail Vladimirovich Belevtsev (Minsk, Belarus)</b>  Deputy Director for Research, Republican Research and Practical Center for Pediatric Oncology, Hematology, and Immunology, Belarus</p>
14.30-14.50	<p>Primary Immunodeficiencies in Pediatric Practice: Clinical Cases  <b>Marina Alekseevna Morenko</b>  Head of the Department of Pediatric Diseases with Courses in Allergology, Hematology, and Endocrinology, NAO "Astana Medical University"  Doctor of Medical Sciences, Professor Chief Pediatric Allergist-Immunologist of the Astana Health Department</p>
14.50 – 15.10	<p>Hereditary Angioedema: A Congenital Immune System Defect – A Multifaceted Healthcare Challenge  <b>Arai Zhanatovna Batyrbayeva (Almaty, Kazakhstan)</b>  Head of the Republican Allergology Center at the JSC "Scientific Research Institute of Cardiology and Internal Diseases," Almaty</p>
15.10 – 15.30	<p>Features of Hematopoietic Stem Cell Transplantation in Patients with Congenital Immune Deficiencies  <b>Lyubov Vyacheslavovna Zherko (Belarus)</b>  Republican Research and Practical Center for Pediatric Oncology, Hematology, and Immunology, Belarus, Transplantologist (<i>Online</i>)</p>
15.30 – 16.00	<p>Primary Immunodeficiencies in Children (Predominantly Antibody Deficiencies, Diagnosis, and Burden)  <b>Mikhail Vladimirovich Belevtsev (Minsk, Belarus)</b>  Deputy Director for Research, Republican Research and Practical Center for Pediatric Oncology, Hematology, and Immunology, Belarus  (<i>Supported by Takeda</i>)</p>
16.00 – 16.20	<b>Coffee break</b>
<b>Section: One Meter Apart</b>	
<b>Moderators: I.Y. Mukatova, E.T. Tuleutayev</b>	
16.20 – 16.40	<p>Cystic Fibrosis: Yesterday, Today, Tomorrow  <b>Elena Lvovna Amelina (Moscow, Russia)</b>  Candidate of Medical Sciences, Senior Researcher at the Cystic Fibrosis Laboratory, FSBI "Research Institute of Pulmonology," Federal Medical and Biological Agency of Russia. Member of the Expert Working Group on Diagnosis of the European Cystic Fibrosis Society (ECFS) (<i>Online</i>)</p>
16.40 – 17.00	<p>Genetics of Cystic Fibrosis: Clinical Aspects  <b>Stanislav Alexandrovich Krasovsky (Moscow, Russia)</b></p>

	Candidate of Medical Sciences, Acting Head of the Cystic Fibrosis Laboratory, FSBI "Research Institute of Pulmonology," Federal Medical and Biological Agency of Russia ( <i>Online</i> )
17.00 – 17.15	Genetic Diagnosis of Cystic Fibrosis in Kazakhstan <b>Asiya Magdatovna Bazenova</b> Geneticist, Clinical and Genetic Diagnostics Department, Corporate Fund "University Medical Center"
17.15 – 17.35	Neonatal Screening for Cystic Fibrosis: International Standards <b>Victoria Davidovna Sherman (Moscow, Russia)</b> Candidate of Medical Sciences, Senior Researcher, Cystic Fibrosis Scientific and Consultation Department, FSBSI "Medical Genetic Research Center named after Academician N.P. Bochkov," Moscow, Russia ( <i>Online</i> )
17.35 – 17.55	Cystic Fibrosis in Children – Challenges, Achievements, and Prospects <b>Tatyana Vasilievna Marshalkina (Almaty, Kazakhstan)</b> Pediatric Pulmonologist Candidate of Medical Sciences, Highest Qualification Category Physician Head Pediatric Pulmonologist of the Ministry of Health of the Republic of Kazakhstan ( <i>Online</i> )
17.55 – 18.15	Organization of Outpatient Management of Adult Patients with Cystic Fibrosis at the Research Institute of Cardiology and Internal Diseases <b>Saule Abubakirkyzy Ungar (Almaty, Kazakhstan)</b> Pulmonologist, Center for Orphan Pathology, Research Institute of Cardiology and Internal Diseases ( <i>Online</i> )
18.15 – 18.35	Targeted Therapy for People with Cystic Fibrosis: International Experience <b>Isabelle Sermet-Gaudelus (France)</b> Professor of Pediatrics, Head of the Cystic Fibrosis Department, National Pediatric Reference Center for Cystic Fibrosis and CFTR-Related Diseases, Paris, France ( <i>Online</i> )
18.35 – 18.55	Cystic Fibrosis and Pregnancy: First Experience in Kazakhstan <b>Gulmira Kurbanovna Rapilbekova</b> Medical Director – Board Member, Corporate Fund "University Medical Center" Doctor of Medical Sciences, Obstetrician-Gynecologist

<b>Day 2</b>	
<b>28.02.2025</b>	
<b>Palliative Care and Rehabilitation</b>	
<i>Session: «Art of care: support, comfort and quality of life in palliative care»</i>	
<b>Moderators: E.V. Polevichenko, N.A. Maltabarova, Sh.A. Bulekbaeva</b>	
08.00-08.30	Palliative Care for Patients with Neuromuscular Diseases <b>Elena Vladimirovna Polevichenko (Moscow, Russia)</b> Doctor of Medical Sciences, Professor of the Department of Oncology, Hematology, and Radiation Therapy, Pediatric Faculty, Pirogov Russian National Research Medical University, Ministry of Health of Russia Head Pediatric Specialist in Palliative Care, Ministry of Health of Russia Board Member of the Association of Professional Hospice Care Providers, Moscow, Russia ( <i>Supported by PTC Therapeutics</i> )



8.30 – 8.50	Rehabilitation: Not All Are the Same <b>Sholpan Adilzhanovna Bulekbaeva</b> Chair of the Board, JSC "Republican Children's Rehabilitation Center" Highest Category Neurologist, Doctor of Medical Sciences, Professor, MBA Head Pediatric Rehabilitation Specialist of the Ministry of Health of the Republic of Kazakhstan
08:50-09:10	Respiratory and Nutritional Support for Children with Neuromuscular Diseases <b>Yulia Leonidovna Streletskaya</b> Pediatric Anesthesiologist-Intensivist, Pediatric Neurologist, Pediatrician ( <i>Online</i> )
09:10-09:30	Experience of Rehabilitation Treatment for a Patient with SMA at the NCDR <b>Aigul Toleubaevna Altynbekova</b> Deputy Chair of the Board for Medical Affairs, NAO "National Center for Children's Rehabilitation" PhD Candidate, I.K. Akhunbaev Kyrgyz State Medical Academy
9.30 – 10.00	<b>Discussion:</b> Analysis of Lethal Cases in SMA. Multidisciplinary Team Work <b>Participants:</b> Dinmukhamed Nurnyazovich Ayaganov, Altynshash Khairullayevna Jaxsybayeva, Bakhytkul Jusupzhanovna Myrzaliyeva, Latina Aizhanovna Tekebayeva
<b>10.00 –10.20</b>	<b>Coffee break.</b>
10.20 – 10.50	Principles of Pain Management in Children with Orphan Diseases <b>Elena Vladimirovna Polevichenko (Moscow, Russia)</b> Doctor of Medical Sciences, Professor of the Department of Oncology, Hematology, and Radiation Therapy, Pediatric Faculty, Pirogov Russian National Research Medical University, Ministry of Health of Russia Head Pediatric Specialist in Palliative Care, Ministry of Health of Russia Board Member of the Association of Professional Hospice Care Providers, Moscow, Russia
10.50 – 11.10	Challenges in the Rehabilitation of Children with Epileptic Encephalopathies <b>Leila Oralgazievna Dairbayeva (Almaty/Astana, Kazakhstan)</b> MBA, Kazakhstan National League Against Epilepsy Center for Neurology, Epileptology, and Rehabilitation Head Epileptologist of Almaty
11.10 – 11.30	Nursing Management in Gene Therapy for Children with Spinal Muscular Atrophy <b>Nurlan Yerzhanovich Terlikbayev</b> Senior Nurse, Pediatric Neurology Department, Corporate Fund "University Medical Center"
11.30 – 11.50	"Family-Centered Care for Neuromuscular Diseases: Principles of Nursing Care" <b>Balaua Rashidkyzy Temirkhan</b> Nurse, Pediatric Neurology Department, Corporate Fund "University Medical Center"
11.50 – 12.10	"Organization of Nursing Care During Immunotherapy for Pediatric Patients with Solid Tumors" <b>Erkin Saginbayuly Nurdjaubaev</b> Nurse, Oncology Department No. 3, Corporate Fund "University Medical Center"
12.10 – 12.30	Challenges in Monitoring Children with Orphan Neurological Diseases in Kazakhstan <b>Latina Aizhanovna Tekebaeva</b> Candidate of Medical Sciences, Head of the Pediatric Neurology Program, Corporate Fund "University Medical Center," National Research Center for Maternal and Child

	Health. Head Pediatric Neurologist of the Ministry of Health of the Republic of Kazakhstan
12.30 – 12.45	Mutations in Ion Channel Genes in Patients with Inherited Forms of Arrhythmias <b>Ainur Rakhmetulovna Akilzhanova</b> MD, PhD, DSci, Professor. Chief Research Scientist, Head of the Laboratory of Genomic and Personalized Medicine, National Laboratory Astana, Nazarbayev University
12.45-13.00	Molecular Genetic Diagnosis of Achondroplasia <b>Lyazzat Kenesovna Nazarova</b> Cytogeneticist, Corporate Fund "University Medical Center"
13.00-13.45	<b>Lunch.</b>
<b>Day 2</b>	
<b>Main Conference Hall</b>	
<b>Session: Health Mosaic: An Interdisciplinary Approach to Orphan Diseases</b>	
<b>Moderators: A.V. Bazarova, S.E. Isakov</b>	
13.45 – 14.05	Multidisciplinary Approach to Managing Achondroplasia in Children: The Role of the Endocrinologist <b>Akmaryl Asylovna Nurbekova (Almaty, Kazakhstan)</b> Doctor of Medical Sciences, Professor of the Department of Endocrinology, Kazakh National Medical University named after S.D. Asfendiyarov
14.05 – 14.25	Pharmacological Treatment of Achondroplasia in Children in the Republic of Kazakhstan <b>Isamdun Kaudunovich Kharamov (Almaty, Kazakhstan)</b> Head of the Pediatric Orthopedics Department, Children's Center "Aksai," Kazakh National Medical University named after S.D. Asfendiyarov Candidate of Medical Sciences, Highest Category Physician
14.25 – 14.45	Nephrotic Cystinosis <b>Svetlana Valentinovna Papizh (Moscow, Russia)</b> Candidate of Medical Sciences, Senior Researcher, Department of Hereditary and Acquired Kidney Diseases named after Prof. M.S. Ignatova, Branch of the Research Clinical Institute of Pediatrics and Pediatric Surgery named after Academician Y.E. Veltischev, Pirogov Russian National Research Medical University, Ministry of Health of Russia (Supported by Recordati Rare Disease) (Online)
14.45 – 15.05	Homozygous Familial Hypercholesterolemia <b>Tatyana Vladimirovna Ivanova-Razumova</b> Director of the Pediatrics Department, Corporate Fund "University Medical Center" Candidate of Medical Sciences, MBA Head Pediatric Cardiologist of the Ministry of Health of the Republic of Kazakhstan
15.05 – 15.25	Primary Hyperoxaluria: From Diagnosis to Treatment <b>Samar Yergalievich Isakov</b> Nephrologist, Head Pediatric Nephrologist of the Astana Health Department (Supported by KFK Medservice Plus)
15.25– 16:00	Inborn Errors of Immunity: From Bench to Bedside <b>Nima Rezaei (Tehran, Iran)</b> Professor of Clinical Immunology and Allergy, Tehran University of Medical Sciences

	Associate Dean of International Affairs, School of Medicine Director of the Global Academic Program (GAP)
16.00 - 16.20	<b>Coffee break.</b>
<i>Day 2.</i> 28.02.2025 <b>Main Conference Hall</b> <b>Moderators: Batpenova G.R., Ivanova-Razumova T.V.,</b>	
16.20 – 16.40	New Horizons in the Treatment of Epidermolysis Bullosa <b>Gulnar Ryskeldieva Batpenova</b> Professor, Honored Worker of the Republic of Kazakhstan President of the Kazakhstan Association of Dermatovenereologists and Dermatocosmetologists. Head Dermatovenereologist of the Astana Health Department Head of the Department of Dermatovenereology and Dermatocosmetology, NAO "Astana Medical University", Doctor of Medical Sciences
16.40 – 17.00	Extracutaneous “Companions” of Epidermolysis Bullosa <b>Natalya Olegovna Tsoy</b> Associate Professor, Department of Dermatovenereology and Dermatocosmetology, NAO "Astana Medical University" PhD, Highest Category Physician Doctor at the "Kobelek Balalar" Charitable Foundation, Republic of Kazakhstan
17.00 – 17.20	Nutritional Deficiency in Children with Epidermolysis Bullosa <b>Elena Georgievna Belonogova</b> Pediatrician, Neurologist, Dietitian, Candidate of Medical Sciences Expert at the "Deti-Babochki" Foundation ( <i>Online</i> )
17.20 – 17.40	Organization of Care for Patients with Genodermatoses in Kazakhstan <b>Ainur Abimullayevna Taubaeva</b> Head of the Inpatient Department, Kazakh Scientific Center of Dermatology and Infectious Diseases. Dermatovenereologist
17.40 – 18.00	Our Experience in the Treatment of Hand Syndactyly in Epidermolysis Bullosa <b>Isamdun Kaudunovich Kharamov</b> Head of the Pediatric Orthopedics Department, Children's Center "Aksai," Kazakh National Medical University named after S.D. Asfendiyarov Candidate of Medical Sciences, Highest Category Physician
<b>Day 2</b> 28.02.2025 <b>Conference Hall 2</b> <b>Session: Genes of Hope: Breakthroughs in Pediatric Oncohematology and Rare Diseases</b> <b>Moderators: Abildinova G. Zh., Bayanova M.F.</b>	
13.45 – 14.05	Genetic Registry of Congenital and Hereditary Pathology "QazGenIs" <b>Gulshara Zhusupovna Abildinova</b> Doctor of Sciences, Professor Chief Freelance Geneticist of the Ministry of Health of the Republic of Kazakhstan
14.05 – 14.25	Genetic Testing for Rare Diseases in Genomics Era - Experience from South Korean Reference Lab. <b>Speaker: Seung Woo Ryu (South Korea)</b>

	Lead Clinical genomics scientist Ph.D. in Biochemistry
14.25 – 14.45	Whole-Genome Studies in the Diagnosis of Orphan Diseases <b>Nikolay Bubeev (Turkey)</b> Sequencing Methods Consultant, Illumina Company
14.45 – 15.05	Osteogenesis Imperfecta: Genetic Variants and Diagnosis <b>Mirgul Fayzullinovna Bayanova</b> Head of the Clinical and Genetic Diagnostics Department, Corporate Fund "University Medical Center". Candidate of Medical Sciences
15.05 – 15.25	Rare Microchromosomal Rearrangements in Orphan Diseases: Clinical and Genetic Analysis of Kazakhstani Patients <b>Alina Ravilievna Girfanova</b> Geneticist, Center for Molecular Medicine
15.25 – 15.45	Modern Methods of Diagnosis and Treatment of Phenylketonuria in the Republic of Kazakhstan <b>Alexandra Vladimirovna Murtazaliyeva (Almaty, Kazakhstan)</b> Geneticist, LLP "Center for Molecular Medicine"
15.45 – 16.00	Tackling rare diseases with Genimics. Learning from a country – wide genomics program <b>Tiago Magalhaes (UAE)</b> Vice President for Bioinformatics M42 ( <i>Online</i> ).
16.00 – 16.20	Coffee break
<b>Дата: 28.02.2025</b> <b>Conference hall 2.</b> <b>Moderators: Kyzdarbekova Zabira, Zhienkulova Gulzhanat.</b>	
16.20 – 16.40	Aspects of Providing Care for Patients with Gaucher Disease in Kazakhstan <b>Gulnara Kaldenovna Abdilova (Almaty, Kazakhstan)</b> Oncologist-Hematologist, Candidate of Medical Sciences Head of the Pediatric Oncology and Hematology Center The National Center for Pediatrics and Pediatric Surgery. ( <i>Online</i> )
16.40-17.00	Organization of medical care for children with hemophilia in Kazakhstan: Issues and Perspectives <b>Manzhova Lyazzat Nurbapaevna (Almaty, Kazakhstan)</b> PhD, Associate Professor, Deputy Chairman of the Board for Scientific and Innovation Activities at JSC "Scientific Center for Pediatrics and Pediatric Surgery," Chief Freelance Pediatric Oncologist and Hematologist of the Ministry of Health of the Republic of Kazakhstan ( <i>online</i> ).
17.00-17.15	Plexiform neurofibromas in children with NF1: New approaches to diagnosis and therapy <b>Zhenkulova Gulzhanat Omarovna</b> Oncologist-Hematologist, PhD, Coordinator of the Day Hospital Department of the KAD Pediatrics at the "University Medical Center" in Astana.
17.15-17.30	Experience with the use of arsenic trioxide in children with acute promyelocytic leukemia <b>Kyzdarbekova Zabira Nurakhmetovna</b> Head of the Hematology Program at the CAD Pediatrics, "University Medical Center" in Astana.
17.30-17.45	Congenital Hemolytic Anemias: Features of Diagnosis and Therapy <b>Abileva Asemgul Bulatovna</b>

	Oncologist-Hematologist, Hematology Program at the CAD Pediatrics, "University Medical Center" in Astana.
17.45-18.00	Features of HSCT in TKI-induced Myeloproliferative Disorders <b>Kusheova Gulshat Nurlibekovna</b> Oncologist-Hematologist, Hematology Program at the KAD Pediatrics, "University Medical Center" in Astana.
<b>Day 2</b> <b>28.02.2025</b> <b>Conference hall 1.</b> <b>Session: "Metabolic Disorders in the Context of Rare Gastroenterological, Endocrinological, and Rheumatological Diseases"</b> <b>Moderators: Kanatova A.K., Sharipova M.N.</b>	
13:45-14:05	"Rare Liver Diseases - Focus on Hereditary Tyrosinemia Type 1" <b>Sharipova Maiya Nabimuratovna (Almaty, Kazakhstan)</b> Doctor of Medical Sciences, Professor, Pediatric Gastroenterologist of the Highest Category at the National Center for Pediatrics and Pediatric Surgery in Almaty.
14:05-14.25	Diagnosis of Lipodystrophy in Children: A Clinical Case <b>LapteV Dmitry Nikitich (Moscow, Russia)</b> Professor of the Russian Academy of Sciences, Doctor of Medical Sciences, Professor of the Department of Pediatric Endocrinology, Head of the Pediatric Department of Diabetes at the Federal State Budgetary Institution "National Medical Research Center of Endocrinology" of the Ministry of Health of the Russian Federation ( <i>online</i> ).
14:25-14:45	Frequency and Structure of Hereditary Metabolic Diseases Based on Selective Screening Results <b>Sklyarov Vladislav Vladimirovich (Almaty, Kazakhstan)</b> Geneticist at the Center for Molecular Medicine.
14:45-15:05	Alagille Syndrome: Diagnosis and Treatment <b>Kanatova Aigul Kanatovna</b> Pediatric Gastroenterologist, Chair of the Public Organization "Pediatric Intestinal Health Protection," Head of the Gastroenterology Department at the "University Medical Center."
15.05-15:25	Osteoporosis in Children: A Rare Diagnosis or an Underestimated Problem? Modern Approaches to Diagnosis and Treatment <b>Rakhimzhanova Marjan Kumanovna</b> Head of the Endocrine and Hereditary Diseases Program, MSPH, Endocrinologist at the "University Medical Center."
15.25-15:45	Alpha-mannosidosis: Etiology, Pathogenesis, Treatment <b>Kanatova Aigul Kanatovna</b> Pediatric Gastroenterologist, Chair of the Public Organization "Pediatric Intestinal Health Protection," Head of the Gastroenterology Department at the "University Medical Center."
15:45 -16.00	Clinical Forms of Hypophosphatasia: Experience in Observing and Long-Term Treatment of Patients with Hypophosphatasia <b>Kalinichenko Natalia Yuryevna (Moscow, Russia)</b> PhD, Senior Researcher at the Federal State Budgetary Institution "MNRC Endocrinology" with support from AstraZeneca ( <i>online</i> ).
16.00 – 16.20	Coffee break

Date: 28.02.2025

Moderators: Asylbekova M.K., Rakhimzhanova M.K.

16.20-16.40	Capillaroscopy in Rheumatological Practice. <b>Speaker: Violetta Opoka-Winiarska (Poland).</b> Medical University of Lublin, Department of Pediatric Pulmonology and Rheumatology, Professor ( <i>online</i> ).
16.40-17.00	Fibrodysplasia Ossificans Progressiva – How to Recognize It? <b>Speaker: Asylbekova Maikesh.Kuantaevna</b> Head of the Rheumatology Program, CF "University Medical Center," Physician of the Highest Category ( <i>with support from IPSEN</i> ).
17.00-17.20	Juvenile Idiopathic arthritis, systemic type, diagnostic and therapy challenges. <b>Speaker: Mukusheva Zaure Serikpaevna</b> Pediatric rheumatologist, Physician of the Highest Category, CF "UMC", Head pediatric rheumatologist.
17:20-17.40	Macrophage Activation Syndrome in Children: A Life-Threatening Condition, Diagnostic Criteria and Treatment <b>Albekova Janara Omerbekovna</b> Resident of the Rheumatology Program at the Department of General Pediatrics, "University Medical Center."
17.40-18.00	Crioperin associated periodical syndrome in children (CAPS), early diagnostic and treatment. <b>Speaker: Dildabaeva Aiken.</b> Resident of the Rheumatology Program at the Department of General Pediatrics, "University Medical Center."
<b>Day 3.</b> <b>01.03.2025</b> <i>Main conference hall.</i> <i>Time: 08.00-18.00</i> <b>Section: Patient's Day</b> <b>Moderators: Ivanova-Razumova T.V., Tuleutaev E.T., Baigalieva B.M</b>	
8.00 – 9.00	<b>Registration.</b>
9.00 – 9.30	Welcome speech. <b>Nurlybaev Erzhan</b> , Vice Minister, Ministry of Health of the Republic of Kazakhstan.
09.30-09.40	Welcome speech. <b>Venera Khanapieva Altynova</b> Deputy Medical Director of the UMC, Director of the Department of Medical and Regulatory Affairs at UMC.
9.40 – 9.50	Welcome Speech <b>Tuleutaev Yernas Tuleutaevich</b> Head of the Republican Center for Rare Diseases, CF UMC, Astana

9.50 – 10.00	"Smart School" Hospital School as Part of an Integrative Approach to the Treatment and Rehabilitation of Children with Rare Diseases.  <b>Elmira Aytashevna Aliyeva</b> – Chair of the "Help Today" Foundation, Member of the Board of Trustees of the "Kazakhstan Halkyna" Foundation, Almaty.
10.00 –10.10	Ensuring Sustainable Provision of Medicines: Priorities. <b>Speaker: Taukeleva Saida</b> Chairperson of the Board of the Public Association "Union of Patient Organizations of Eurasia", Almaty.
10.10 –10.30	Coffee Break
<b>Nervous System Diseases</b> <b>Moderators: Tuleutaev E. T., Tekebaeva L. A., Syzdykova B. R.</b>	
10.30-10.40	Ataxia and Huntington's disease. <b>Speaker: Anarbek Urkumbaev</b> Founder of the Public Association "Senim," Astana.
10.40-10.50	The role of the "Omirge Sen" Foundation in supporting patients with neuromuscular diseases. <b>Speaker: Sabiya Shugelova</b> Director of the "Omirge Sen" Foundation, Almaty.
10.50-11.00	Orphan genetic syndromes, including epilepsy: diagnosis, treatment, and monitoring. <b>Speaker: Svetlana Kirichkova</b> Director of the "Ken Zhurek" Foundation, Almaty.
11.00-11.10	Myasthenia. Advances in diagnosis, new treatment methods, and challenges faced by patients. Congenital myasthenic syndromes – "a lost nosology". <b>Speaker: Imanbekov Anuar</b> Founder and head of the informal association of patients with Myasthenia Gravis in Kazakhstan.
11.10-11.20	Issues related to ALS (Amyotrophic Lateral Sclerosis). <b>Speaker: Aidymbaeva Akzhamal</b> Founder of the "Life with ALS Matters" Foundation.
11.20-11.30	Fund's work results and current status of patients with Rett syndrome. <b>Speaker: Roza Berkesheva</b> Director of the "Erekeshe Zhol" Foundation.
11.30-11.40	Rett syndrome – an orphan disease, methods of assistance and treatment. International experience. <b>Speaker: Rustam Kadiyev</b> President of the "Zhan-Bota" Children's Foundation.
11.40-11.50	"The Path to Health for SMA Patients in Uzbekistan" Online.  <b>Vladislav Kochetkov</b> – Founder of the patient organization for children with SMA in Uzbekistan "SMA Oilasi," under the Republican Fund for the Social Adaptation of Children.
11.50 – 12.20	Discussion

**Endocrine System Diseases, Eating Disorders, and Metabolic Disorders  
Diseases of the Digestive System**

**Moderators: Tuleutaev E. T., Sharipova M. N., Abduakhasova G. Kh., Kanatova A. K.,  
Durmanova A. K., Rakhimzhanova M. K.**

12.20-12.30	Support and development of the "Zhan Omir" Foundation <b>Speaker: Ramazanov Samat</b> Director of the "Zhan Omir" Foundation, Almaty.
12.30-12.40	Diagnosis and treatment of MODY diabetes in Kazakhstan: challenges and prospects. <b>Speaker: Zakiev Ruslan</b> Director of the "DiaRKom" Foundation, Almaty.
12.40-12.50	Difficulties in treating DLDCL. <b>Speaker: Aizhan Abisheva</b> Mother of a child with DLDCL, Almaty.
12.50-13.00	Adrenal insufficiency and how to live with it? <b>Speaker: Nazgul Zhurmagambetova</b> Parental community "HARMONiA," Kostanay.
13.00-13.30	<b>Coffee break</b>
13.30-13.40	Issues in diagnosing and treating children with glycogen storage disease. Parents' requests? <b>Speaker: Gulzhan Khudaibergenova</b> Parental community for glycogen storage disease, Astana.
13.40-13.50	Congenital hyperinsulinism in Kazakhstan. <b>Speaker: Maksut Zhientaev</b> Father of a child with CHI, Kostanay.
13.50 – 14.00	Cystic Fibrosis today: main challenges of patients in RK. <b>Speaker: Nurmagambetova Gaukhar</b> Parental community of patients with cystic fibrosis.
14.00 – 14.10	Drug Provision <b>Speaker: Dusmanbetov Ardak</b> Member of the Board of Trustees of the Public Fund for Assistance and Support of People with Inflammatory Bowel Diseases, Almaty.
14.10 – 14.20	Phenylketonuria <b>Speaker: Saida Imamuddinova</b> Representative of the Parents' Community.
14.20 – 14.50	Discussion
<b>Congenital Anomalies (Developmental Defects), Deformations, and Chromosomal Disorders</b>	
<b>Moderators: Tuleutaev Ernas, Taubaeva A., Rahymzhanova Marzhan, Durmanova Aigul.</b>	
14.50-15.00	"Butterfly Wings": Uniting Efforts for the Future <b>Speaker: Zhylkybai Aly</b> Director of BF "OF Kobelek Balalar".
15.00-15.10	The Role of NGOs in Improving the Quality of Life for People with Achondroplasia. <b>Speaker: Svetlana Abdulova</b> Director of OF "Small World," Karaganda.



15.10-15.20	"Mastocytosis: A Difficult Path to Diagnosis and the Fight for Patient Safety in a Limited Healthcare System". <b>Speaker: Elena Vogonova</b> , Mother of a Child with Mastocytosis, Almaty.
15.20-15.50	<b>Discussion</b>
<b>Circulatory System Diseases</b> <b>Diseases of the Blood, Hematopoietic Organs, and Immune System Disorders</b> <b>Moderators: Tuleutaev E. T., Ivanova-Razumova T. V., Mukarov M. A., Kyzdarbekova Z. N.</b>	
15:50-16:00	Pulmonary Hypertension and Lung Transplantation. <b>Speaker: Alla Koroleva</b> , Director of OF "New Breath," Almaty.
16:00-16:10	Key Goals and Objectives of the Kazakhstan Association of Hemophilia Patients with Disabilities. <b>Speaker: Akzhan Erikkyzy</b> President of the Kazakhstan Association of Hemophilia Patients with Disabilities, Almaty.
16:10-16:20	Women with Bleeding Disorders. <b>Speaker: Lyudmila Shagabutdinova</b> , Regional Coordinator of the Association of Hemophilia Patients with Disabilities, Almaty.
16:20-16:50	<b>Discussion</b>
<b>Diseases of the Musculoskeletal System and Connective Tissue</b> <b>Moderators: Tuleutaev E. T., Asylbekova M. K., Aitzhanova R. M.</b>	
16:50-17:00	Life with Vasculitis: Vera, Nadezhda, Lyubov. <b>Speaker: Elmira Khusainova</b> , Director of the Fund "Together with Takasu," Almaty.
17:00-17:10	Pregnancy Planning with SLE (Systemic Lupus Erythematosus): How to Reduce Risks? <b>Speaker: Raushan Moldabekova</b> , Project Coordinator of OF "Lupus Kazakhstan," Almaty.
17.10 – 17.40	<b>Discussion.</b>
17:40-18:00	Public Association "Association for Assistance to Patients with Rare Diseases in the Republic of Kazakhstan" <b>Closing Remarks</b>  <b>Speaker: Baigaliyeva Bakhyt Madenietovna</b> <ul style="list-style-type: none"> <li>• Founder of the Public Foundation “Charity Fund ‘Kobelek Balalar’”</li> <li>• Founder and Head of the Association of Legal Entities “Association for Assistance to Patients with Orphan Diseases in the Republic of Kazakhstan”</li> <li>• Member of the Board of Trustees of the Public Foundation “Qazaqstan Halqyna”</li> <li>• Member of the Local Commission on Bioethics of NAO “Astana International University”</li> </ul>