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9th World Congress On

Rare Diseases and Orphan Drugs

June 17-18, 2019
Berlin, Germany

S C I E N T I F I C P R O G R A M

08:30-09:00 **Registrations**

09:00-09:30 **Introduction**

09:30-09:50 COFFEE BREAK

09:50-11:50
Meeting Hall

KEYNOTE LECTURES

MEETING HALL

Rare Diseases in Neurology
Rare Mental and Behavioral disorders
Rare Pediatric Diseases
Immunological Rare Diseases
Rare Diseases in Nephrology

13:10-13:15 GROUP PHOTO

13:15-14:00 LUNCH BREAK

MEETING HALL

Rare Skin Disorders
Rare Oncology
Rare Diseases of Lymphatic System
Rare Nutritional and Metabolic Disorders

16:00-16:20 COFFEE BREAK

MEETING HALL (16:20-17:00)

**Young Researchers in
Rare Diseases and Orphan Drugs**

MEETING HALL (17:00-18:00)

Workshop

09:00-10:30
Meeting Hall

KEYNOTE LECTURES

10:30-10:50 COFFEE BREAK

MEETING HALL

10:50-12:50

- Rare Gynecological and Obstetrical diseases
- Treatment and Advanced Therapies options for Rare Diseases
- Orphan drugs Potentiality
- Clinical case studies on Rare Diseases

12:50-13:35 LUNCH BREAK

MEETING HALL

- Rare diseases in Bloodstream
- Rare diseases of Endocrine System
- Rare Musculoskeletal Diseases
- Rare Hereditary diseases

15:55-16:15 COFFEE BREAK

MEETING HALL (16:15-17:00)

Poster Presentations

MEETING HALL (17:00-18:00)

Workshop

Awards & Closing Ceremony



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**9th World Congress on
Rare Diseases and
Orphan Drugs**

*Theme: Explore the Recent advancements in Rare
Diseases Research*

June 17-18, 2019 | Berlin, Germany

AGENDA

Title: Nitisinone in the treatment of Alkaptonuria

**Lakshminarayan
Ranganath**
Royal Liverpool University
Hospital, UK

Alkaptonuria (AKU) is an iconic autosomal recessive severe multisystem disorder of the tyrosine degradation pathway due to lack of homogentisate dioxygenase resulting in increased circulating and urinary homogentisic acid. Morbidity includes lithiasis (renal, salivary, prostate, gall bladder), osteopenia, fractures, ruptures of ligaments/muscle/tendons, spine and joint disease. An approach to treating AKU by inhibiting the production of HGA by using nitisinone has been recently recognized. Nitisinone has been used in a related tyrosine disorder, hereditary tyrosinaemia 1 (fatal in early childhood) as the standard of care for more than 20 years. This presentation discusses the efforts of our group in developing nitisinone for AKU, an approach consistent with repurposing. Nitisinone is being developed as a licensed therapy in DevelopAKUre, a European Union funded clinical programme. In parallel, nitisinone is also being used off license in a centre (NAC) commissioned by NHS England Highly Specialised Services since 2012. Data collected from the NAC shows a beneficial effect of nitisinone in AKU.

Title: The Effect of G. Lucidum on the Lifespan of Caenorhabditis Elegans modeling Duchenne Muscular Dystrophy

Prashanthi Rayapati
STEM Researcher,
USA

Duchenne muscular dystrophy (DMD) is an X chromosome-linked disease characterized by progressive physical disability, immobility, and premature death in affected boys. Underlying the devastating symptoms of DMD is the loss of dystrophin, a structural protein that connects the extracellular matrix to the cell cytoskeleton and provides protection against contraction-induced damage in muscle cells, leading to chronic peripheral inflammation. However, dystrophin is expressed in neurons within specific brain regions, including the hippocampus, a structure associated with learning and memory formation. Linked to this, a subset of boys with DMD exhibit progressing cognitive dysfunction, with deficits in verbal, short-term, and working memory. into the peripheral effects of dystrophin deficiency.

Title: Red blood cell-encapsulated enzymes: an innovative therapeutic approach to overcome challenges of enzyme replacement therapies for rare diseases

Emmanuelle Dufour
ERYTECH PHARMA,
USA

Many inborn errors of metabolism (IEM) disorders are due to defects in single genes encoding key metabolic enzymes. In most cases, clinical manifestations of these disorders are driven by the over-abundance of a metabolite (the enzyme's substrate) or the scarcity of an essential metabolite (the enzyme's product). Though rare, IEM disorders can have devastating consequences for patients and their families. While some Enzyme Replacement Therapies (ERT) are commercially available for a few IEM disorders, the clinical benefits of these approaches are often outweighed by the emergence of hypersensitivity and the rapid clearance of enzymes. Therefore, there is a high need for better tolerated and longer-acting replacement enzymatic activity to alleviate the burden of IEM disorders.

SPEAKER SLOTS AVAILABLE

Title: The Economics and Sustainability of Orphan Drugs

Carina Schey
University of Groningen,
Switzerland

Orphan drugs are increasingly under scrutiny by reimbursement bodies in Europe. This is due in part to the unexpected rise in the number of orphan drugs that received marketing authorization since 2000. The high prices of some orphan drugs and the budget constraints, affordability and sustainability of access to orphan drugs further contribute to the sometimes negative spotlight on orphan drugs. However, one of the limitations in the current reimbursement pathways is the use of cost-effectiveness analyses to assist in the decision-making process. Increasingly, payers and policy makers highlight the need for alternative methods of assessing the value of orphan drugs and demonstrating their ongoing accessibility. A novel approach, using multicriteria decision analysis, was developed to review orphan drugs. The framework has been tested with useful results.

Title: New therapies in genetic skeletal diseases achieved through drug repurposing

Michael D Briggs
Newcastle University,
UK

Genetic skeletal diseases (GSDs) are an extremely diverse and complex group of diseases that primarily affect the development and homeostasis of the skeleton. There are more than 450 unique and well-characterised phenotypes that range in severity from relatively mild to severe and lethal forms and although individually rare, as a group of related orphan diseases, GSDs have an overall prevalence of at least 1 per 4,000 children, which represents a large unmet medical need. Our studies have focussed on a group of clinically-related GSDs that present with disproportionate short stature and early onset OA and result from dominant-negative mutations in a range of cartilage structural proteins including cartilage oligomeric matrix protein (COMP), matrilin-3, aggrecan and types II, IX and X collagens.

Title: Empowering and Engaging HIV+ women 50+ through collective dialogue to address stigma and discrimination: An ICW-North America

Chantal Mukandoli
ICWNA,
Canada

Background: There is a strong association between HIV-infection and sexually transmitted infections (STIs) in MSM all over the world. The goal of our research is to determine the prevalence of gonorrhoea, chlamydial infection, M. genitalium-infection, and syphilis among HIV-positive MSM. **Methods:** 381 MSM living with HIV were recruited through clinics and non-governmental organizations. To evaluate the prevalence of STIs three probes from each patient were collected: first void urine (FVU), pharyngeal and rectal swabs (PS and RS). The samples were tested for DNA of N. gonorrhoeae, C. trachomatis, M. genitalium, Herpes Simplex Virus (HSV) I and II, T. pallidum in PCR. **Results:** The prevalence of STDs in FVU: N. gonorrhoeae – 1.6% (6/381), C. trachomatis – 3.7% (14/381), M. genitalium – 1.3% (5/381), T. pallidum – 0.5% (2/381). In RS: N. gonorrhoeae – 11.3% (43/381), C. trachomatis – 18.4% (70/381), M. genitalium – 4.7% (18/381), T. pallidum – 3.4% (13/381). In PS: N. gonorrhoeae – 6.8%

SPEAKER SLOTS AVAILABLE

Title: Rare oral soft tissue metastasis: an overview.

Cinzia Casu
AIRO,
Italy

Metastasis to the oral cavity is a rare event and constitutes 1% of all oral cavity malignancies. We would like to report the cases in the literature of rare oral soft tissue metastasis. We reported oral metastasis on the cheeks, on the lips, on the floor of the mouth, on soft palate and on the uvula. We had found only 8 cases of oral metastasis on the lips searching on PubMed with the keywords “oral metastasis in lips”, “lip metastasis”. The primary malignant tumor that most frequently metastasizes at the labial level is certainly the renal tumor with 5 cases out of 8. In 5 cases the upper lip was affected, there was 1 case in which metastasis was present in both lips, and this is a very interesting data because the other malignant diseases are more frequent in the lower lip.

Title: Primary Hydatidosis of distal femur masquerading malignancy – A rare case.

Rajni Prasad
VMHC and Safdarjung Hospital

Hydatidosis of bone is a very rare condition and its incidence as compared to other organs range from 0.5-4%. There is no typical clinical appearance and the image characteristics on Xray and CT are similar to those of tuberculosis, metastasis, giant cell tumour or bone cyst. Case Summary- A 53 year old female presented to the orthopaedics OPD of Safdarjung hospital with pain and swelling over her right knee region. Routine radiograph exhibited multiple lytic lesions with thinned out cortices and fracture of supracodylar region. A provisional clinical diagnosis of malignancy was made and fine needle aspiration was advised. Cytology was reported as suspicious of malignancy and histopathology was asked for confirmation. Biopsy was reported as Hydatid disease of bone. **Conclusion:** Early diagnosis is uncommon in primary hydatidosis of bone as it is not considered in the differential diagnosis of osteolytic lesion.

Title: CHANGE IN GONADOTROPINS IN POSTMENOPAUSAL WOMEN: EFFECTS OF PARITY

Ekhatior C.N
St Philomena Catholic Hospital

Abstract: This report assessed the effect of parity on gonadotropins pattern in postmenopausal women. We studied 280 post menopausal women (40 each grouped into nuliparous to para 6). Although there was no significant difference in their ages, serum follicle stimulating hormone (FSH) and luteinizing hormone (LH) appeared to correlate negatively with parity. Thus, the levels of gonadotropins may vary with parity in postmenopausal women. **Results:** The mean age of the women ranges from 56.05±6.91 to 59.25±5.45 years. **Discussion :** Based on the results, serum gonadotropin levels may vary with parity as with age, BMI, lifestyle and ethnicity.

SPEAKER SLOTS AVAILABLE

Title: Clinical Case of Congenital Hyperinsulism

N.B. Belykh
Russia

Relevance. Congenital hyperinsulinism (CHI) is a rare hereditary disease characterized by insulin hypersecretion and severe persistent hypoglycemia in children. The aim of the study is to present a clinical case of CHI in a child born of mother with type 2 diabetes. **Patients and methods.** Analysis of the clinical case and medical documentation. **Results.** The girl from the 3rd pregnancy proceeding against the background of type 2 diabetes, 3 preterm births (35–36 weeks), weigh is 3410 g. After birth, the glycaemia was 0.1 mmol/l and then stabilized (5.0–4.3 mmol/l). In the first year of life glycaemia was in the range of 3.0–4.0 mmol/l, the neurodevelopment corresponded to the age. At 11 months of age, the level of insulin was 17.4 µE/ml. At the age of 1 year on the background of a long hungry pause glycaemia was 1.6 mmol/l; the child became lethargic, convulsions were noted.

Title: Wyburn-Mason syndrome. Ever heard of it?

Dan Jeffries
Patient, Author and Advocate
UK

Dan Jeffries has, and his insightful and entertaining talk explores what it's like living with one of the world's rarest medical conditions - and then finding out you have another one. Dan was diagnosed with Wyburn-Mason syndrome at four-years old. It is an exceptionally rare AVM (Arteriovenous Malformation), situated around the midbrain and optic nerve. As a consequence, Dan is blind in his left eye. The AVM is untreatable and there are thought to be less than 100 reported cases in the past fifty years — worldwide. That works out to be about 1 in 70 million people. Pretty rare. “Dan’s talk managed to be informative and entertaining, insightful and moving, educational and inspiring. You don’t often find presentations that manage to combine all of those qualities! I left with a deeper understanding of what it is like to live with rare conditions, and also with a smile on my face!”

Title: Lysosomal Storage Disorders - Updates from India

Shashank Tyagi,
Lysosomal Storage Disorder
Support Society

Lysosomal Storage Disorders Support Society (LSDSS) a first, not-for-profit and pan India organization is registered and headquartered in New Delhi, India, headed by a group of parents and patients of LSD who know what it’s like to live with Lysosomal Storage Disorders. LSDSS was formed formally in year 2010 to address the unmet needs and create awareness that benefit patients with lysosomal disorders in India. LSDSS works for creating awareness for prevention, timely and accurate diagnosis, consultation, symptomatic care and treatment options amongst patients, physician’s, care givers and hospitals across India. LSDSS is a prime force behind the formation of National Rare Disease Policy in India by approaching the Judiciary through legal cases. LSDSS is doing advocacy in Government of India at Central and State level for better implementation of National Rare Disease policy.

SPEAKER SLOTS AVAILABLE

Title: Non-dystrophic myotonic disorders: Patients' Insights on Treatment Access

Carina Schey
University of Groningen,
The Netherlands

Background: Non-dystrophic myotonic (NDM) disorders are a heterogeneous group of rare, genetic skeletal muscle chloride and sodium channelopathies with altered membrane excitability resulting in prolonged muscle contraction and delayed relaxation. Patients experience stiffness, pain, weakness, impaired mobility, fatigue, disability, falls, problems with speech, chewing and swallowing. **Method:** A two-stage study ascertaining the level of access to mexiletine and benefits for those treated with mexiletine was developed and conducted in required languages. **Results:** Online questionnaires were completed by 37 NDM patients, of whom 41% were currently taking mexiletine. Of those not taking mexiletine, 67% had never heard of mexiletine and 25% reported it not being available in their country. **Conclusion:** Mexiletine-treated patients experienced substantial benefits, while denied access resulted in substantial harm.

Title: Prevalence of Urinary Tract Infections and Risk Factors among Pregnant Women attending ante natal Clinics in Government Primary Health Care Centres in Akure

O.J Odeyemi
Federal University of
Technology

Urinary tract infection has become the most common bacterial infections in humans, both at the community and hospital settings, it has been reported in all age groups and in both sexes. This study was carried out in order to determine and evaluate the prevalence, current drug susceptibility pattern of the isolated organisms and identify the associated risk factors of UTIs among the pregnant women in Akure, Ondo State, Nigeria. A cross-sectional study was conducted on the urine of pregnant women and socio-demographic information of the women were collected. A total of 300 clean midstream urine samples were collected and a general urine microscopic examination and culture were carried out, Microbact identification system was used to identify gram negative bacteria.

Title: Pandemic-The effectiveness of vaccination in urban medicine

**Rishat
Ahmetvaleev**
Russian Federation

The pandemic is near. Alberta Health Services says the second wave of influenza affects the southern zone and this is a different strain. Until mid-January, the A1 H1N1 flu was the dominant strain in Alberta this season. Vivienne Sutthorpe, medical specialist for AHS in the southern zone. Low flu vaccine effectiveness Among all people at risk, the effectiveness of vaccination in urban medicine, according to first estimates, is "moderate" (59%) against the A (H1N1) pdm09 virus and "weak" (19%) against influenza A (H3N2), says Sp France. These preliminary results were obtained from the Sentinel network, which evaluates the effectiveness of the vaccine to prevent influenza infection leading to consultation with a general practitioner, explains the health agency.

SPEAKER SLOTS AVAILABLE

Title: HIV + women’s current knowledge and access to community services by ICWNA

Chantal Mukandoli
 ICWNA
 Canada

Women living with HIV who are 50 years of age or older (HIV+ Women who are 50+) are facing stigma while disclosing their status to their family, friends, community, and partners. As a result, HIV+ women are frequently facing issues that negatively impact their mental health, such as depression, isolation, violence, and trauma. The principles of greater involvement and meaningful engagement (GIPA/MIPA) would require that HIV+ women are empowering and educating other HIV+ women so they too can become leaders in the community. Diverse community supports create empowering spaces for HIV+ women to address barriers currently preventing their adherence to medications, access to services, and further HIV education. Specifically, for HIV+ women over 50 these spaces enable women to maintain their inherent dignity, safety.

Title: Rapid progression osteolysis in Gorham Stout Syndrome. A case report and literature review

Leonardo Cano Cevallos
 Universidad Catolica
 Santiago De Guayaquil

Background: Gorham-Stout Syndrome is a rare idiopathic nonmalignant disorder characterized by recurrent, progressive osteolysis. It may affect any bone, but commonly involves mandible, shoulder and pelvic girdle, each in roughly 20% of all the cases. The disease affects one or contiguous bones. **Case presentation:** A 12-year-old-boy is admitted by complete loss of function of the right arm and showing a deformity at level of right clavicle. His history was characterized by progressive weakness and pain on right shoulder 4 months ago. **Conclusion:** The time of progression of the disease in a normal pattern is at least one or two years within the beginning of suggestive symptoms. We consider that our case is particularly important because the progression to total destruction of elements of shoulder’s patient was four months in a young patient.

SPEAKER SLOTS AVAILABLE

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