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Analysis of patient access to orphan/rare drugs in Pakistan

Rizwan Arshad
Pakistan Public Health Association, Pakistan

Background: Orphan/rare diseases are chronic, serious, and life-threatening conditions that have a low prevalence and require complex care. Worldwide, as estimation one in 15 people are affected by rare diseases. This study aims to analyze the accessibility, reimbursement status, licensed status of drugs and __ (ATC) codes of drugs that the European Medicines Agency (EMA) in Pakistan considers to be "orphan" pharmaceuticals.

Methods: The drugs included in this analysis were obtained from the list of orphan drugs published by the EMA. Orphan drugs' accessibility and licensing status in Pakistan were obtained from the list of FDA orphan drugs. Descriptive analysis was applied to determine the accessibility status of orphan drugs identified by the EMA in Pakistan.

Results: Based on the EMA, 105 pharmaceuticals were approved with "orphan drug" status except for drugs that have lost orphan drug status, decommissioned in the European Union and withdrawn from the European Community Register by January 2020. Of the 105 rare drugs on the EMA list, 84 were inaccessible in Pakistan. Of the 21 available drugs, 1 (0.87%) were licensed and 20 (99%) were unlicensed in Pakistan. Among 1 (0.87%) of licensed products and 20 (99%) of unlicensed products were not covered by reimbursement. When orphan drugs' ATC codes were examined, the most common ATC group was found to be "L—Antineoplastic and Immunomodulatory" agents.

Conclusion: An orphan drug incentive policy is very important to ensure early access to the drugs used to treat orphan/rare diseases. Considering the capacity and prices for orphan drugs in Pakistan, it can be said that almost every patient with orphan/rare diseases have difficulty in their treatment. It is obvious that such a policy must prepare for the regulation of orphan drugs in Pakistan.

Keywords: Rare disease drug, Orphan drug, Orphan drug market

Biography

Rizwan Arshad is a young public health researcher having 13 years of experience on accessibility of medicines (ATM) primarily hard to hard to find medicines including rare and orphan drugs for the vulnerable patients in Pakistan. Currently he is working on the model development of one window services model (OWSM) for the accessibility of cancer immunotherapies and orphan/rare drugs on named patient basis. It is his research model and core interest area as well. He has been associated with many organizations as a consultant for the alignment of health services along with regulatory authorities and identification of gaps in health for the communities and provides the solutions thereof. He is also responsible for making an outreach to peripheries within the country for the accessibility of quality drugs to the community people.

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Detection of viral RNA in conjunctival secretions of hospitalized patients with confirmed Covid-19 infection

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Lahore General Hospital, Pakistan

To detect the presence of viral RNA in conjunctival swabs of patients with positive naso-pharyngeal swabs for Covid-19, Cross sectional study in Covid Admission Unit & EYE UNIT 1, Lahore General Hospital, from November 2020 to April 2021 (06 months).Approval was taken from Ethical Review Board of LGH by obtained from all patients we have taken Patients of both gender from the age 18 years & above. Positive PCR test from nasopharyngeal swab for Covid-19 criteria based on Patients mechanical ventilation we have taken patients topical eye drugs conjunctival swabs were collected from both eyes by the same ophthalmologist using PPE and done with LGH laboratory performed the analysis for all swabs in the statistical analysis data was entered and analyzed using SPSS v25.0 and Chi-Square test was applied to see the correlation between Conj. swabs and Serum markers and resulted A p-value ≤0.05 was considered significant .Conjunctive swabs collected from 66 patients that is 132 eyes and Covid-19 RNA was detected in 06 patients (9.1%), some other results Conjunctiva swabs Positive with Ocular Symptoms = 50% and Without Ocular Symptom = 6% considering P-Value (Chi-Square = 0.002) Highly Significant. Final Conclusions would be viral RNA for Covid-19 can be detected in app. 9% of Conj. Swabs and Positive Conj. swab is strongly associated with the presence of occular symptoms and such patients often have normal values of serum markers like D-Dimer, Ferritin, CRP & LDH.

Biography

Ghazala Rubi is working in Molecular Genetics Research Lab. this is the Central Research Lab of Post Graduate Medical Institute, Lahore General Hospital. This Lab caters all facilities of Molecular & Genetics to all Researchers of Post Graduate Medical Institute and post graduates research Projects.

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Lacrimal sac lymphomas in pediatric age group

Dalal R Fatani* and Hailah AL Hussein
Lahore General Hospital, Pakistan

14-year-old boy was referred to King Khaled Eye Specialist Hospital as case of left nasolacrimal duct obstruction for Dacryocystorhinostomy. The child complained of left eye tearing for one year duration followed by localized swelling over the medial canthal for one month duration. The mas firm with bony consistency and caused no proptosis. The lacrimal system was clearly obstructed and the probe could not be passed. The patient then underwent CT scan which highlighted a well-defined 8.2 x 13.1 mm lesion filling the left nasolacrimal sac with extension inferiorly into a dilated nasolacrimal duct. The left nasolacrimal duct measured 7 mm in diameter while the right unaffected side measured 4.5 mm.

Surgical exploration was carried out revealing a gelatinous fleshy mass filling the entire lacrimal sac and incisional biopsy was taken. The biopsy revealed a lacrimal sac lymphoma.

The lacrimal sac is lined by a pseudostratified columnar epithelium with cilia and goblet cells. Lacrimal sac tumors are divided into epithelial and non-epithelial tumors based on the tumor origin. 60-94% of lacrimal sac tumors are classified under epithelial tumors. Benign epithelial tumors include Papillomas which are the most common comprising 36%, oncocytomas and adenomas. Malignant epithelial tumors in the lacrimal sac are squamous cell carcinomas, oncocytic adenocarcinoma, mucoepidermoid carcinoma and cystic adenoid carcinoma. Inverted papillomas may be considered premalignant lesions and they are associated with human papilloma virus 6 and 18 alongside with squamous cell carcinoma.

Lacrimal sac lymphomas are categorized as lymphoproliferative non epithelial malignant tumors of the lacrimal sac. They are rare with a median age of onset at 51 years. Most common type is mucosa-associated lymphoid tissue (MALT) or diffuse large B cell lymphoma (DLBCL). Lacrimal sac lymphoma in pediatric age group is very rare. In a review of 250 pediatric patients with orbital tumors in YEAR by Shields et al, only 6 cases had orbital lymphoma none of which were within the lacrimal sac. Few cases have been reported in the literature after this review was published.

Biography

Dalal R Fatani is an aspiring ophthalmologist aiming to continue my career growth and further expand my knowledge, surgical skills, and research contributions in the field. She is member of the king Saud medical city training unit to promote resident's wellbeing and facilitate their journey. She is Producer and host of an educational podcast directed towards ophthalmology residents. Certified Suba diving (advanced level diver), Mountain climbing (local hiking excursions, Mount Kilimanjaro for Animal rights and welfare, Mount Shams in Oman for GCC fitness), Animal rescue activities with Open Paws Riyadh, and Reading (self-improvement and parenting books).

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Does fever increase or decrease blood circulation?

K M Yacob
Marma Health Centre, India

This is the first time many people have heard such a question.

When it comes to treating back pain, neck pain, and knee pain, it is often heard that the cause of the pain is reduced blood flow. A variety of heat-inducing devices are used to increase blood flow to the lower back, neck, and knee pains. Physiotherapy often provides more heat than fever.

1. What happens to blood flow in your body when your internal temperature decreases?

Vasoconstriction, Thermogenesis, when there is a decrease in blood flow and its signs, symptoms, and signals, the immune system does actions to increase blood flow to save lives.

2. What are the ingredients necessary to decrease blood flow?

The main ingredient is the lack of enough temperature. Shrinking of blood vessels, Inflammation, infection, low pressure, etc... will decrease blood flow.

3. Decreased blood flow can cause fits, delirium, stroke, and lead to death.

When disease increases essential blood circulation and energy levels also decrease. The vertical height between the heart and brain is more than one foot. When the disease becomes severe, the ability to pump blood to the brain decreases. Then blood flow to the brain decreases and delirious or fits are formed. As a result of this brain cells are damaged. So the patient might be paralyzed or may even die. 87% of stroke is due to blood to the brain is decreased or blocked.

4. What is the Purpose of the temperature of a fever?

When the disease made by the bacteria, fungi, venom, horror scene, horror dream, etc..., becomes a threat to life or organs blood circulation decreases, the temperature of fever will emerge to increase prevailing essential blood circulation. And it acts as a protective covering of the body to sustain life.

There is no way other than this to increase prevailing essential blood circulation for a sensible and discreet immune system to protect the life or organ.

In all diseases which decrease essential blood circulation and temperature, the fever will emerge to Increase essential blood circulation and temperature. Fever is an adaptation and a result of Thermogenesis. To this day, no one has heard that fever is caused by poor blood flow.

5. What are the ingredients necessary to increase blood flow?

Adequate temperature and pressure, free flow of blood, and disease-free condition are all factors that increase blood flow.

6. The temperature of the fever increases the blood flow.

Fever increases blood flow, which means more lymphocytes flow through lymphoid tissues. The body temperature of the brooding hen increases to provide the required temperature for the egg and to increase the essential blood circulation in the body. The brooding hen does not eat anything. The increased temperature is its food. It helps to convert fat into energy. Similarly, your immune system generates fever to increase the body's essential blood circulation.

7. Will fever cause decreased blood flow? Or damage brain cells?

No. Fainting or delirious or damage to the brain cells is not due to the increased temperature of fever. It is due to a decrease in blood circulation to the brain. Temperatures below 42 degrees never cause any harm to our bodies.

During summer, in some parts of India atmospheric temperature is more than 45 degrees. While taking a normal Steam bath the temperature inside the box is more than 50-degree centigrade, Physiotherapy Treatment temperature is between 52.5 °C to 54.4 °C. There is no such a history of one having fainted, or being affected with delirious or fits.

The fever is heat energy. To date, modern science has not studied what actions were carried out heat on fever.

The cause of all complications, including death, is the

treatment of fever without knowing why it is hot.

What kind of treatment should be given if you have symptoms of decreased blood flow?

Treatment should be to increase blood flow.

This is the basic principle of physics.

Is there any benefit in reducing body heat during fever?

There is no merit of any kind.

Not only is it of no benefit, but it also causes death by inflammation and infection.

8. Ways to eliminate fever by increasing blood circulation.

The actual treatment for fever is to increase blood circulation. Two ways to increase blood circulation

1. To increase essential blood circulation never allow body temperature to lose via the atmosphere and
2. Apply heat from outside to the body. When the temperature produced by the body due to fever and heat which we applied to the body combined together, the blood circulation increases.

A hot Bathtub, Steam bath, hot sandbag or thermal heat pad, hairdryer, blanket, Hot pepper water, and hot drinks can be used to increase blood circulation. A way to get heat all over the body from outside the body to increase blood circulation.

8.1. Setup a hot water bathtub. Always set its temperature at 42 degrees celsius with the help of a thermostat. If a person with has to fever is submerged in water from the bottom of the nose to the point where the soles of the feet are completely submerged. This will help all parts of the body will receive the same amount of heat.

9. How can we prove that the temperature of fever is to increase essential blood circulation?

9.1. It has been proven around the world that all types of heat increase blood flow.

A variety of heat-inducing devices are used to increase blood flow to the lower back, neck, and knee pain. Physiotherapy often provides more temperature than fever. Physiotherapy Treatment temperature is between 52.5 °C to 54.4 °C.

9.2. There is no fundamental difference between the temperature used in physiotherapy and the temperature of the fever.

9.3. If we measure the thermal energy used for any activity in fever, we can understand how temperature increases blood flow and what the purpose of heat in fever is.

9.4. As the temperature rises, the free flow time of blood, erythrocytes, and plasma decreases.

9.5. Shivering is to increase blood circulation. It is a part of Thermogenesis.

Heat-reducing fever treatment with water and paracetamol should be banned as soon as possible.

Biography

K M Yacob is a practicing physician in the field of healthcare in the state of Kerala in India for the last 34 years and very much interested in basic research. He interest is spread across the fever, inflammation and back pain. He is a writer. He already printed and published ten books on these subjects. He wrote hundreds of articles in various magazines. After scientific studies, we have developed 8000 affirmative cross checking questions. It can explain all queries related to fever.

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Behaviors of probiotic formulations in aromatic fixed and essential oils and established therapeutic models: In vivo studies of repair of dysbiosis with coconut oil and trace peppermint-lemon-tefarik essential oils and multiprobiotic replacement

Hulya Kayhan
Art de Huile, Turkey

It is known that dysbiosis occurs with the decrease of beneficial bacteria, increase in pathogens and decrease in bacterial diversity. The microbiota in the healthy host, the homeostatic conditions of all organisms, benefit host development and health. However, some important factors such as antibiotic use or unhealthy diet can disrupt the structure of the microbial community. These disruptions can result in the loss of microorganisms that are beneficial to the host. There is increasing evidence that dysbiosis of the gut microbiota is associated with the pathogenesis of both intestinal and extra intestinal disorders. Inflammatory bowel disease, irritable bowel syndrome (IBS) and Small Intestinal Bacterial Overgrowth (SIBO) have the most important place among the intra-intestinal disorders, while metabolic disease tables such as allergy, cardiovascular disease deficiencies and obesity take place at the beginning of the extraintestinal disorders [2]. SIBO is a syndrome characterized by an increased number of abnormal bacterial species in the small intestine. It is also defined as the clinical and laboratory representation of the relationship between qualitative and/or quantitative changes of the gut microbiota and malabsorption. The duodenum and proximal jejunum normally contain less than 10E+4 CFU per mL of microorganisms. In this study; The therapeutic effects of probiotic bacteria including Akkermancia muciniphila and Lactobasillus gastricus combined with aromatic fixed and essential oils on SIBO will be shared using the "Dysbiosis-Based Rat SIBO (Small Intestine Bacterial Overgrowth) Model".

Biography

Hulya Kayhan is Co-Founder of Art de Huile Pharmacist in Aromatherapy. She was Graduated from Faculty of Pharmacy, Istanbul , Turkey. She gives training on aromatherapies in Worldwide Conferences.

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The effects and issues affecting rare patients in Africa due to inadequate access to drugs or medical trials

Nthabeleng Ramoeli
Rare Disease Lesotho Association, Lesotho

The plight of rare diseases patients or Rare Butterflies as we call them in Africa is set against the backdrop of a continent dealing with socio-political and economic conditions that adversely affect vulnerable groups such as rare patients in need of specialist care. The condition of the continent as a whole leaves a gaping hole where those most vulnerable are susceptible to falling through the cracks, this is why non-profit organizations such as Rare Diseases Lesotho Association and the Rare Diseases Initiative exist. These entities exist to drive an active mandate to ensure that everyone, especially Rare patients receive adequate healthcare services and support, while maintaining a firm stance in the promotion of their equitable rights to medical care with dignity at the forefront. This study focuses on the inadequacies and shortfalls of the systems that exist in Africa in reference to rare patients receiving specialist care, accessing lifesaving medicine and potentially life altering medical trials as part of their treatment plans. It is no surprise that the continent as a whole is grossly unprepared and overwhelmed with the need for specialist care facilities and lifesaving orphan drugs and treatment plans, and this sets us back a couple of years in the healthcare system and seek to address these issues through a pragmatic problem solving outlook on the issues illustrated within the scope of access to healthcare for Rare patients and the red tape that exists within.

Biography

Nthabeleng P Ramoeli is the founder of Rare Disease Lesotho Association which was founded in October 2017. She is a young woman with Ehlers–Danlos Syndromes (EDS) and Seeng Nchai, a mother with child with a rare disease. The purpose of the Organization assists all patients affected by rare diseases to access treatment and support care for improved health and quality of life..

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SP1-binding site polymorphism in the COL1A1 gene and its relation to osteoporosis in Egyptian patients with Gaucher disease

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Gaucher disease (GD) is a recessive disorder due to a mutation of the glucocerebrosidase gene leading to reduction in β-glucocerebrosidase activity, accumulation of glucosylceramide. Enzyme replacement is the most common therapy; however, bone manifestations can be slow to respond. COL1A1 is one of the principal candidate genes for fragility fractures in osteoporosis. This study examined the distribution of the polymorphic variant in the regulatory region of COLIA1 gene at a recognition site for transcription factor in Egyptian patients with Gaucher Disease compared to a control group. Thirty Egyptian patients with GD from About El Riche Hospital, Cairo University and 30 healthy age and sex-matched individuals from Fayoum University Hospitals were included as controls. Clinical examination was done for both patients and controls. SP1 binding site polymorphic variant at COL1A1 gene was detected by PCR-RFLP for both groups. Over sixty six percent (66.6%) of the GD patients had the wild genotype (G/G) while 26.7% were heterozygous for G/T polymorphism. Only 6.7% harbored the homozygous T/T variant. The findings make the hypothesis of an association between Sp1 COL1A1 gene polymorphism and bone disease in GD probably feasible and that component strongly influence bone remodeling.

Key words: COL1A1 gene, Gaucher disease (GD), Bone Mineral Density (BMD), Osteoporosis, osteopenia.

Biography

Shahira Elshafie had completed her MD Degree of Clinical and Chemical Pathology in Kasr El-Aini Medical School Cairo University, 1996. She had received the Basic Certificate of Medical Education from Cairo University in 2016. She is the head of Department of Clinical and Chemical Pathology, Fayoum University from 2016. She had received the Best Paper Award, Initial Steps for Internationalization of Medical Education in Egypt and International Business and Education Conference, Las Vegas, 2014. She is the Egyptian Syndicate of Medical Doctors, since 1984 and Founding member of the Women in Higher Education for Leadership, 2011.

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What does a technology enabled patient concierge mean to the orphan drugs industry?

Harsha K Rajasimha
Jeeva Informatics Solutions Inc., USA

Traditional clinical research paradigms relying solely on brick and mortar in-person engagement between researchers and patients have failed to provide the scale and efficiencies needed in orphan products development. Patients with rare disease are geographically sparsely distributed globally, are already burdened by the disease they carry, often genetic and debilitating, are often dependent on a caregiver to take paid time off to support them, yet are still willing to travel across the globe to access a life-saving or altering treatment options such as gene therapies. But it is unfair to expect them to do so when it is really not necessary in this day and age of Telemedicine, Digital Health, and wearable devices. Unique to rare diseases R&D are patient registries and natural history studies. These multi-year studies are often necessary prerequisites for orphan product development as patients demonstrate significant heterogeneity of symptoms with limited medical literature and understanding of their physiological and molecular underpinnings. The traditional process of patient education, engagement, informed consent, screening, enrollment, retention, and evidence generation needs an overhaul. It is not just introducing digital tools and elements into the centralized brick-and-mortar paradigm. It is about finding and meeting patients where they are in the global communities through online social channels and multi-modal engagement. I will demonstrate how such as technology enabled patient concierge is helping connect patients with registries, natural history studies, clinical trials, other global resources and experts based on their specific needs.

Biography

Harsha K Rajasimha is a precision medicine data scientist by training with 17+ years of experience spanning academia, the National Institutes of Health, FDA, healthcare and life science consulting, and multiple startups. He is a social entrepreneur focused on accelerating the research and development of diagnostics and therapies for rare and common diseases. He has founded numerous international organizations to address these challenges. He is pioneering human-centric technology innovation to accelerate online recruitment of diverse patients for clinical research in a range of diseases including rare, chronic, infectious, and neurobehavioral disorders. He has authored 17+ publications, book chapters, and patents.

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The whole centered approach - from the lens of a "professional patient"

Deborah Vick
RareABILITY, USA

Patient Centered Care or the "whole" patient approach has been growing in private and public practices worldwide. Empowering patients allows the caregivers, patients and their medical team to collaboratively create individualized health care plans that are mutually beneficial to all parties. Patient centered programs place the patient at the head of the table while affording medical team members, caregivers, holistic practitioners and wellness facilitators a seat at the table. A variety of studies have documented the positive benefit and unique characteristics of the "whole patient centered" approach on one's overall health and wellbeing. This discussion will provide a brief overview of the key points from previous research while taking a deeper dive into the development and implementation of said programs. There will be interactive discussions as to what roles patient led advocacy, advocacy organizations and medical professional advocacy play in the creation, development and implementation into patient centered care.

Biography

Deborah Vick is the CEO and Co-founder of Rare ability. She is a mother, educator, a Rare Disease Advocate, Professional Patient and Keynote Speaker. She is mindfulness and meditation facilitator as well as a sound bath practitioner. She is an author and active community volunteer. Community volunteerism includes afterschool STEAM programs, serving as Ms. Wheelchair California USA 2022 and serving as volunteer advisory member of The Magical Bridge Foundation, an organization that helps create, implement and consult on the creation of inclusive playgrounds. In addition, she is an active moderator on several social media and auditory apps in which she helps facilitate daily talks on variety of topics including health and wellness, sustainability leadership and empowering the disability communities.

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