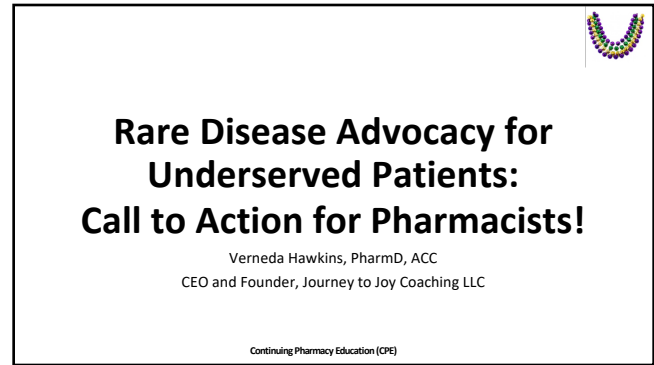




1



2

Learning Objectives

- Define the term "rare diseases"
- List at least 3 examples of rare diseases that disproportionately impact underserved patients
- Summarize the Orphan Drug Act of 1983, the 21st Century Cures Act and other relevant policies that support patients with rare diseases
- Identify common barriers to care for patients with rare diseases to include additional hurdles specific to underserved patients
- Formulate an action plan for pharmacists to collaborate and advocate to eliminate racial healthcare disparities within the rare disease community

Continuing Pharmacy Education (CPE)

3

Disclosures

- None

Continuing Pharmacy Education (CPE)

4

What is the definition of a rare disease?

- USA classifies a disease as being rare when less than 200,000 people are affected (about 1 in 1500 people)
- Rare diseases are estimated to affect over 30 million people in the USA
- About 80% of rare diseases have a genetic component (primarily pediatric patients)
- Definition of a rare disease varies globally
 - Incentivizes drug development based on regional prevalence
 - For ex...In EMA (Europe): affects no more than 1 person in 2000
 - Reference list of global definitions for rare disease <https://www.keionline.org/wp-content/uploads/KEI-Briefing-Note-2020-4-Defining-Rare-Diseases.pdf>

Continuing Pharmacy Education (CPE)

5

What defines an Orphan Drug in the USA?


- A drug intended to treat a condition affecting fewer than 200, 000 persons in the USA
- OR
- A drug which will not be profitable within 7 years following approval by the FDA

fda.gov

Continuing Pharmacy Education (CPE)

6

Orphan Drug Act of 1983 Turns 40!



- Provides incentives for research into rare diseases & possible treatments
 - Grants to provide funding for clinical trials & natural history studies
 - Tax credits for qualified clinical trials
 - Potential seven years of market exclusivity after approval
 - Exemptions from user fees
- Prior to 1983, only 38 drugs were approved to treat rare diseases
- Since its passage, more than 7000 rare diseases have been identified and over 1100 orphan indications for treatments have obtained FDA approval.
- Fewer than 10% of the vast number of rare diseases have a treatment and/or a cure

Continuing Pharmacy Education (CPE)

7

2012 FDA Safety & Innovation Act (FDASIA, PDUFA V)




- Prescription Drug User Fee Act (PDUFA) (launched 1992)
 - Addressed lag in FDA review/approval time (avg time was 29 months!)
 - Mandates metrics for FDA to review & approve/reject marketing applications within 10 months
 - FDA collects fees from the pharma industry
 - Reauthorized every 5 years with enhancements
- FDASIA, 2012 (examples of benefits for rare disease patients)
 - Established a pediatric rare disease priority review voucher
 - Enhanced patient focused drug development

Continuing Pharmacy Education (CPE)

8

21st Century Cures Act (2016)



- Designed to help accelerate medical product development and bring new innovations/advances who need them faster & more efficiently. Examples include:
 - Improves FDA's ability to recruit & retain staff to keep updated on the latest science
 - Enhances FDA's scientific tools to review drugs
 - Increased funding to FDA and NIH
 - Compassionate Use reform
 - Patient focused drug development improvements

Continuing Pharmacy Education (CPE)

9

2023 Congressional Omnibus Spending Act (Public Law 117-328)



- Requires diversity action plans for clinical trials used by the FDA to decide whether drugs are safe and effective
 - Note: Excludes orphan drugs

Continuing Pharmacy Education (CPE)

10

Knowledge Check 1



Which statement is false?

- The USA classifies a disease as rare when less than 200, 000 people are affected
- In the USA, rare diseases are estimated to affect over 30 million people
- A majority of rare diseases have a genetic component
- The global definition for rare diseases is exactly the same.
- All of the above statements are true.

Continuing Pharmacy Education (CPE)

11

Knowledge Check 2



Which of the following legislation initially established a pediatric priority review voucher for rare diseases?

- Orphan Drug Act of 1983
- Congressional Omnibus Spending Bill (Public Law 117-328) (2023)
- 21st Century Cures Act (2016)
- Food & Drug Administration Safety & Innovation Act (PDUFA V) (2012)

Continuing Pharmacy Education (CPE)

12

General Barriers to Rare Disease Patient Care



- Clinical evidence is based on small numbers of patients
 - Limited incidence and outcome data
 - Limited resources i.e funds, patients to undertake a clinical trial
- Finding an appropriate medical expert is challenging which results in extensive patient evaluations to characterize the specific diseases
- Access and involvement of multiple specialties warrants coordination & collaboration
- Significant burden (emotionally, physically, financially) on patients, families & caregivers who are often the best source of information

Continuing Pharmacy Education (CPE)

13

Disparities Amongst Underserved Patients with Rare Diseases



- Rare Diseases vary in prevalence between populations. Examples impacting Black Americans include:
 - Sickle cell disease
 - Sarcoidosis
 - Focal segmental glomerulosclerosis (FSGS)
- FDA reports only 9% of rare disease clinical trial participants are Black which can be attributed to:
 - Lack of awareness
 - Education
 - Inadequate commitment to recruit diverse populations

Continuing Pharmacy Education (CPE)

14

Disparities Amongst Underserved Patients with Rare Diseases (Group Brainstorming Activity)



What are other potential challenges to consider for historically underrepresented patients (and their caregivers/families) in the rare disease community?

Continuing Pharmacy Education (CPE)

15

Sickle Cell Anemia (SCD)



- SCD is an inherited disease marked by defective hemoglobin.
- SCD affects approximately 100,000 Americans
 - Occurs in about 1/365 Black/African American Births
 - Occurs in about 1/16,300 Hispanic-American births
- Patients w/ SCD experience chronic acute pain & vaso-occlusive crisis
- Unfortunately, bias & stigma still exists for patients with SCD
- Need to reinforce HCP care points
 - Listen and believe SCD pain reports
 - Avoid stigmatizing language when referring to SCD and their pain
 - Be aware of your own and others implicit bias

Continuing Pharmacy Education (CPE)

16

Sarcoidosis



- Sarcoidosis is an inflammatory disease of unknown cause characterized by the formation of granulomas in one or more organs of the body.
 - Overdrive of immune system can result in interference of organ's structure and function, primarily the lungs, due to chronic inflammation and fibrosis
- Black Americans, especially women, :
 - have the highest incidence of sarcoidosis in the US
 - have the highest hospitalizations & mortality rates of all groups
 - are more than twice as likely to have a family member with sarcoidosis
 - experience more complications from side effects from treatments to manage sarcoidosis i.e. higher cumulative doses of steroids
 - RDDC 2021 Webinar on Blacks Women & Sarcoidosis: <https://youtu.be/T-G1D2ONnys>

Continuing Pharmacy Education (CPE)

17

Sarcoidosis



- To reduce health disparities, the Foundation for Sarcoidosis Research (FSR) launched 2 initiatives as a call to action for patients and providers:
 - **Ignore No More: African American Women & Sarcoidosis Campaign (2021)**
 - Educates the broader community on how to improve diagnosis, treatment and outcomes
 - **Ignore No More: ACTe Now! (Advancing Clinical Trial for Equity in Sarcoidosis Campaign (2022)**
 - Focuses on a specific pathway Black Americans can take via clinical trial participation to improve care & treatment

Continuing Pharmacy Education (CPE)

18

Focal Segmental Glomerulosclerosis (FSGS)



- **FSGS** : rare type of kidney disease that causes scarring in the filters of the kidneys
- **FSGS** can be caused by a variety of conditions to include diabetes, SCD, other kidney diseases, obesity, drug toxicity, infections i.e. HIV or of unknown origin.
- **FSGS** can also be caused by inherited abnormal genes i.e G1 and G2 renal-risk variants in the apolipoprotein L1 gene (APO1). Recently discovered to be more common in Blacks than Whites primarily due to prevalence of these genetic variants among those of African ancestry.
 - RDDC 2021 Webinar: Black and Rare Understanding APOL1: <https://youtu.be/WL32mfy5rhc>
- Many Black American patients with non-diabetic etiologies of ESKD remain at risk to be mislabeled as having kidney disease attributed to effects of hypertension or other non genetic causes due to the misunderstanding of this disease mechanism.

Continuing Pharmacy Education (CPE)

19

DEI Efforts in Rare Disease Organizations



Inaugural NORD/RDDC Survey: 2022 Key Findings

- Data from 127 individual responses (representing at least 38 unique organizations) received from June-September 2022
- Results from the survey and key informant interviews indicated:
 - Lack of knowledge of how to incorporate DEI concepts within framework
 - Identified DEI as an add on program rather than intrinsic within their organizations
- Examples of recommendations for resolution include:
 - Create DEI goals & objectives within organization's strategy to include outcome measures
 - Engage proactively in targeted recruitment efforts for underserved patients at project/research trial launch
 - Specific personnel training on how to sensitively & thoroughly collect & utilize demographic information

Continuing Pharmacy Education (CPE)

20

Knowledge Check 3



Which of the following statements are **TRUE** regarding sarcoidosis?

- Black American **men** with sarcoidosis have the highest prevalence and greatest burden when compared to all other groups impacted by sarcoidosis in the US.
- Sarcoidosis is an inflammatory disease characterized by the formation of tiny clumps of inflammatory cells that can interfere with an organ's structure and function.
- Sarcoidosis remains difficult to diagnose, with limited treatment options and no known cure.
- Only b and c are true
- a, b and c are true

Continuing Pharmacy Education (CPE)

21

Call to Action: Rare Disease Health Care Team



- Stay up to date on emerging research & legislation on disparities in care and treatment
- Educate your patients about clinical trials and explain the benefits of participation
- Be an effective advocate and partner for your patients by helping to improve the quality of provider/patient communication
 - *Ask and actively listen to your patient about their lives and any challenges they and their caregivers may have about understanding and following their treatment plan.*
- Be self-aware and take a personal inventory to ensure you (and other members of the HCP team) are identifying and addressing biases to better serve your patients

Continuing Pharmacy Education (CPE)

22

Call to Action: Role of the Pharmacist



- **Educate** patients/caregivers on medication to include dose preparation and administration
- **Recognize** when to report and/or address side effects that can signal disease progression and/or treatment failure
 - If applicable, incorporate pharmacogenomics ,defined as the application of genetic information to predict an individual's response to therapy ,to improve efficacy and minimize untoward medication effects
- **Collaborate** with patients, caregivers and healthcare team , as appropriate, to assess availability of patient assistance programs to address medication costs.

Continuing Pharmacy Education (CPE)

23

Call to Action: Pharmacist Education



- Be familiar with rare diseases that are most prevalent in underserved patients <https://everylifefoundation.org/rare->
- Complete CE programs on rare diseases ex. Powerpak, CME/ACPE presentations
 - ask/submit questions and/or provide feedback by recommending incorporation of DEI within training curriculum on evaluation forms
- Attend health equity educational conferences/summits (virtual/in-person/archived)
 - <https://globalgenes.org/event/rare-health-equity-forum/>
- Stay up to date on relevant legislation that impacts underserved patients in the rare disease community (ex. fda.gov, CE)

Continuing Pharmacy Education (CPE)

24

Call to Action: Pharmacist Volunteer Opportunities



- Organizations (nonprofit)
 - **Disease specific** (examples):
 - **SCA**: American Sickle Cell Association
 - **Sarcoidosis**: Foundation for Sarcoidosis Research (FSR)
 - **FSGS**: NephCure
 - **NORD** (National Organization for Rare Disorders) <https://rarediseases.org/>
 - **Global Genes** <https://globalgenes.org/>
 - **RDDC** (Rare Disease Diversity Coalition) <https://www.rarediversity.org/>

Continuing Pharmacy Education (CPE)

25

Call to Action: Future Pharmacist Pipeline



- Inform students, interns and early career pharmacists (0-5 years experience) of relevant fellowships and scholarships/grants designed to build a pipeline of much needed underrepresented HCPs in the rare disease community.
- Examples: NORD and RDDC offers:
 - Internships
 - Fellowships
 - Research Grant
 - Health Equity Conference Attendance Scholarships
 - Note: RDDC application due date is August 31, 2023

Continuing Pharmacy Education (CPE)

26

Call to Action: Pharmacist Career Opportunities



- **Pharmaceutical Industry**
 - Field Medical Liaison (MSL)
 - Drug Information
 - Headquarters Medical Director/Manager
 - Patient Advocacy
 - Research/Clinical Development
 - Commercial/Marketing

Continuing Pharmacy Education (CPE)

27

Call to Action: Pharmacist Career Opportunities



- **Clinical/Academia**
 - Pediatric/High risk Obstetrics
 - IM/FP/Ambulatory/Community
 - Specialty
 - Nephrology: Kidney
 - Heme/Oncology i.e SCA, multiple myeloma
- **Non-profit organizations/Government**
 - Director/Associate Director/Manager
 - Public Health/Health Equity Project Lead

Continuing Pharmacy Education (CPE)

28

Summary



- For underserved populations, challenges are more pronounced compared to other patient groups with rare diseases
- Paucity of underrepresented patients in clinical trials, especially in genome wide associating studies, results in a lack of understanding and subsequently access to effective treatments.
- Pharmacists can play an essential role as a member of the health care team as **"the medication expert"** and a **"patient advocate"** in our quest to reduce disparities for underserved patients and their families/caregivers in the rare disease community.

Continuing Pharmacy Education (CPE)

29

Next Steps: Action Plan for Pharmacists




- Ask yourself/reflect.. then implement...

"What is at least one (1) action step that I can take in my pharmacy practice moving forward to help close the healthcare disparity gap for underserved patients with a rare disease?"

Continuing Pharmacy Education (CPE)

30



Question and Answer

Continuing Pharmacy Education (CPE)

31



Thank You!

Verneda Hawkins, Pharm.D., ACC
CEO & Founder, Journey to Joy Coaching LLC
info@j2jcoaching.com

Continuing Pharmacy Education (CPE)

32