“The ‘Eyes’ Have It!”
A Review of Rare Diseases

Mindy Burnworth, PharmD, FASHP, FAzPA, BCPS
Midwestern University College of Pharmacy-Glendale
Professor of Pharmacy Practice
mburnw@midwestern.edu

Volunteer Ambassador Arizona Rare Action Network
(powered by the National Organization for Rare Disorders)
melinda.burnworth@rareaction.org
is seeing believing

: visual illusions can distort our perception so that what we “see” does not correspond with what is physically there

Aye
“Yes”

Nays
“No”
Learning Objectives

① Distinguish rare diseases and orphan drugs from common diseases and drugs

② Compare and contrast rare disease resources

③ Given a rare disease scenario, formulate a treatment plan
Distinguish rare diseases and orphan drugs from common diseases and drugs

“A rare disease is defined as one affecting < 200K persons.”
-NORD, CORD, EURORDIS, GARD

“50% of people affected by rare diseases are children.”
-NORD, CORD, EURORDIS, GARD

“Over 7,000 distinct types of rare diseases exist.”
-NORD, CORD, EURORDIS, GARD

“Patients with rare diseases are frequently misdiagnosed or undiagnosed.”
-NORD, CORD, EURORDIS, GARD

“Only 5% of rare diseases have an FDA-approved treatment.”
https://www.youtube.com/watch?v=eeVSyYQI4h0&feature=youtu.be
-NORD, CORD, EURORDIS, GARD

“Families and private foundations provide ~3% of ALL medical research funding in the US.”
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“General knowledge & awareness of rare diseases is often gained ‘on the job’ rather than in the required curriculum.”
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“Precision medicine is focusing on the one individual.”
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“Alone we are rare. Together we are strong.”
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② Compare and contrast rare disease resources

rare diseases are not so rare
We Are All Zebras: How Rare Disease Is Shaping the Future of Healthcare

When you hear hoofbeats, think of horses, not zebras." The famous medical maxim in the 1800s was less a hint about rare diseases. That was then. This is now. How rare is precision medicine, a revolution in healthcare based on the rare disease model. Precision medicine uses the unique all of us and focuses not on what makes you part of the herd but what makes you unique.

What Is Precision Medicine?

- Personalized care, tailored to your needs
- Genomic data helps predict and prevent disease
- Risk-based interventions

We Are Not Average

- Not everyone responds the same way to treatment
- Understanding the genetic makeup of a patient helps
- Dosing of certain medications can be improved

Genetics, Proteins, Metabolism, Oh My!

- Precision medicine requires a holistic approach
- Understanding how genetics and proteins work together
- Metabolism plays a role in how medicines are processed

Zebras Unite!

- Spread the word by sharing this infographic
- Be vocal and passionate
- Be proud of your stripes

Find out what you can do: [http://www.sireninteractive.com/zebras/]

The President’s Precision Medicine Initiative

Obama set aside $215 million and asked a panel of experts to contribute ideas on genetics, biological samples, and clinical trials to help the National Institutes of Health (NIH), identify new targets for treatment and prevention, test the impact of new drugs and therapies, and lay the foundation for precision medicine for many different diseases.

What About the Cost?

The US spends more per person on healthcare than any other developed country and yet our health outcomes are among the worst. But precision medicine may help costs go down and care improve by eliminating waste, avoiding treatments that won’t work, and reducing chronic illness.

Rare Informs the Common

By definition, each rare disease affects less than 200,000 people. But there are 7000 rare diseases and together they have a huge impact. Discoveries in rare diseases like familial hypercholesterolemia lead to knowledge that can be applied to more general conditions, like high cholesterol.

Powered by Patients

Precision medicine pulls patients at the center, providing them with personalized care. It relies on empowered partners. We those who share our stories in rare disease. Welcome to the future of healthcare. By scholaring and collaborating, we’ll make things better for all.

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About GARD

The Genetic and Rare Diseases Information Center (GARD) is a program of the National Center for Advancing Translational Sciences (NCATS) and is funded by two parts of the National Institutes of Health (NIH): NCATS and the National Human Genome Research Institute (NHGRI). GARD provides the public with access to current, reliable, and easy-to-understand information about rare or genetic diseases in English or Spanish.

www.rarediseases.info.nih.gov/GARD
Tools and Resources

- **The Rare Disease Database**
  Search NORD’s Database for reports on more than 1,200 diseases. [Search database](#).

- **Patient & Caregiver Resource Center**
  Find free webinars, fact sheets, infographics and other helpful materials. [View resources](#).

- **Member Resources**
  Resources to help you start or grow your patient organization. [View resources](#).

- **Rare Action Network®**
  Visit our new platform where rare disease patients can fundraise, communicate and advocate. [Start now](#).

- **Patient Assistance Program**
  NORD Patient Assistance Programs help patients obtain life-saving or life-sustaining medication. [View programs](#).

- **Rare Disease Day**
  The international advocacy day to bring widespread recognition of rare diseases as a global health challenge. [Visit site](#).

www.rarediseases.org

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The portal for rare diseases and orphan drugs

"Rare diseases are rare, but rare disease patients are numerous"

Access our Services

- Inventory, classification and encyclopaedia of rare diseases, with genes involved
- Inventory of orphan drugs
- Directory of patient organisations
- Directory of professionals and institutions
- Directory of expert centres
- Directory of medical laboratories providing diagnostic tests
- Directory of ongoing research projects, clinical trials, registries and biobanks
- Collection of thematic reports, Orphanet Reports Series

www.orpha.net
Given a rare disease scenario, formulate a treatment plan.

Porphyria is a well-defined genetic disorder of heme biosynthesis.

Yes, pink-purple urine is a classic symptom of porphyria!
What are signs and symptoms of porphyria?

1. Pain (abdominal)
2. Polyneuropathy
3. Psychological symptoms
4. Pink urine
5. Precipitated by drugs

- Women of reproductive age

Drug database

Search again

1 records found

Search again

This information is not intended as a substitute for medical professional help or advice but is to be used only as a reference.

Definitions
OK = very likely to be safe for prolonged use by individuals with an acute porphyria, based on consistent evidence
OK? = probably safe for prolonged use by individuals with an acute porphyria, based on evidence that is either inconsistent or insufficient to be conclusive
BAU = probably unsafe for prolonged use by individuals with an acute porphyria, based on evidence that is either inconsistent or insufficient to be conclusive
BAD = very likely to be unsafe for prolonged use by individuals with an acute porphyria, based on consistent evidence
NO INFO = evidence insufficient for deriving any conclusion

Search again

1 records found

Search again

https://www.accessdata.fda.gov/scripts/opdlisting/oopd/listResult.cfm
panhematin is FDA-approved for treatment of porphyria

Yes, panhematin is supposed to be rust colored!

Yes, the patient will be blue! a classic sign of methemoglobinemia is cyanosis

Yes, the patient will be blue!
methylene blue is FDA-approved for treatment of methemoglobinemia

Yes, the antidote is supposed to be bright blue!

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