

MINI MED SCHOOL: FINDING THE NEEDLE IN THE HAYSTACK – A JOURNEY THROUGH RARE DISEASE DISCOVERY

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Conflicts

- No conflicts to disclose



Learning Objectives

- Explain what a rare disease is and why diagnosing rare diseases is so important
- Describe the role of a clinician-scientist
- Define inborn errors of immunity and their clinical features



What is a rare disease?

- Statistically: A condition that impacts fewer than 1 in 2,000 people
- Often misdiagnosed or not recognized by medical community
- Patients and their families may face obstacles to care



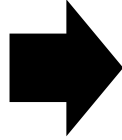
Diagnosing rare diseases can be hard!



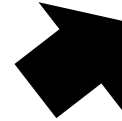
Symptoms develop



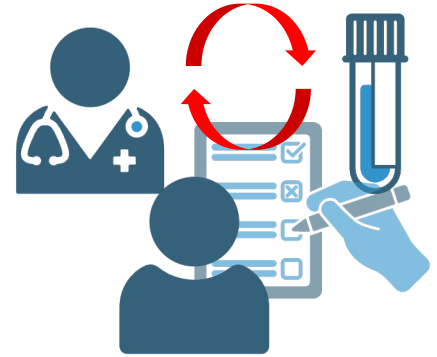
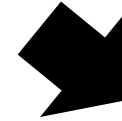
See primary care doctor
Undergo tests



No diagnosis or
suspected diagnosis



Treat symptoms



Refer to other specialists
Order more tests

**Diagnostic journey of a
rare disease patient**





Symptoms develop



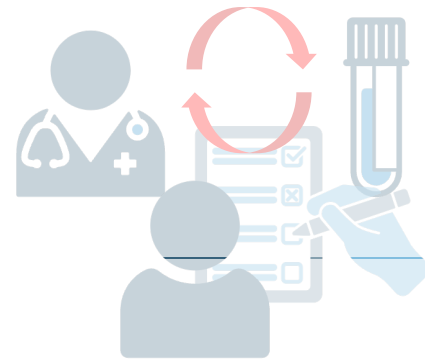
See primary care doctor
Undergo tests



No diagnosis or
suspected diagnosis



Treat symptoms



Refer to other specialists
Order more tests

Specialized testing

- Genetic testing
- Biomarkers



+/- enroll in research studies



Symptoms develop



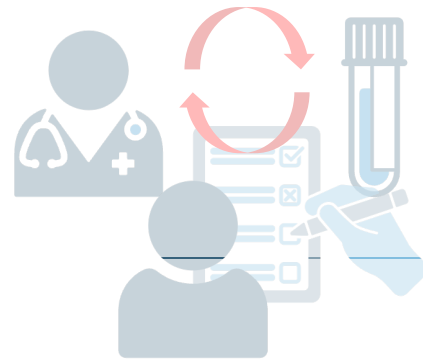
See primary care doctor
Undergo tests



No diagnosis or
suspected diagnosis



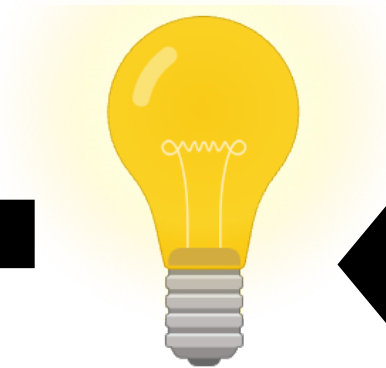
Treat symptoms



Refer to other specialists
Order more tests



Specialized testing



Correct diagnosis!



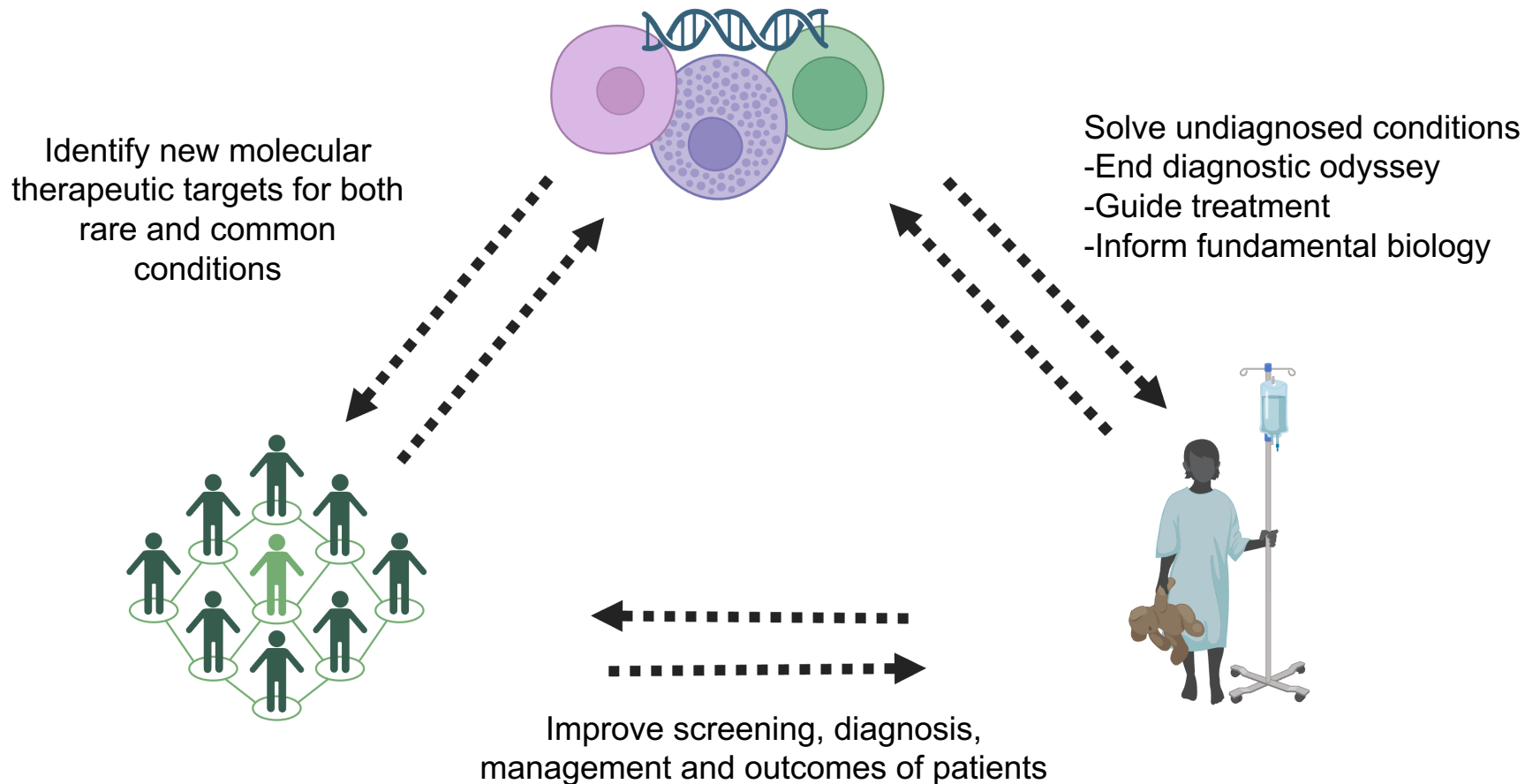
Personalized care

Why is diagnosing rare diseases so important?

- Provides closure to affected patient and family
- Insight re: what to expect in the future
- Opportunity for targeted therapy



Research is critical to rare diseases clinical care!



What is a clinician-scientist?

- Clinician (ex: physician) who splits their time between caring for patients and performing scientific research



Clinic



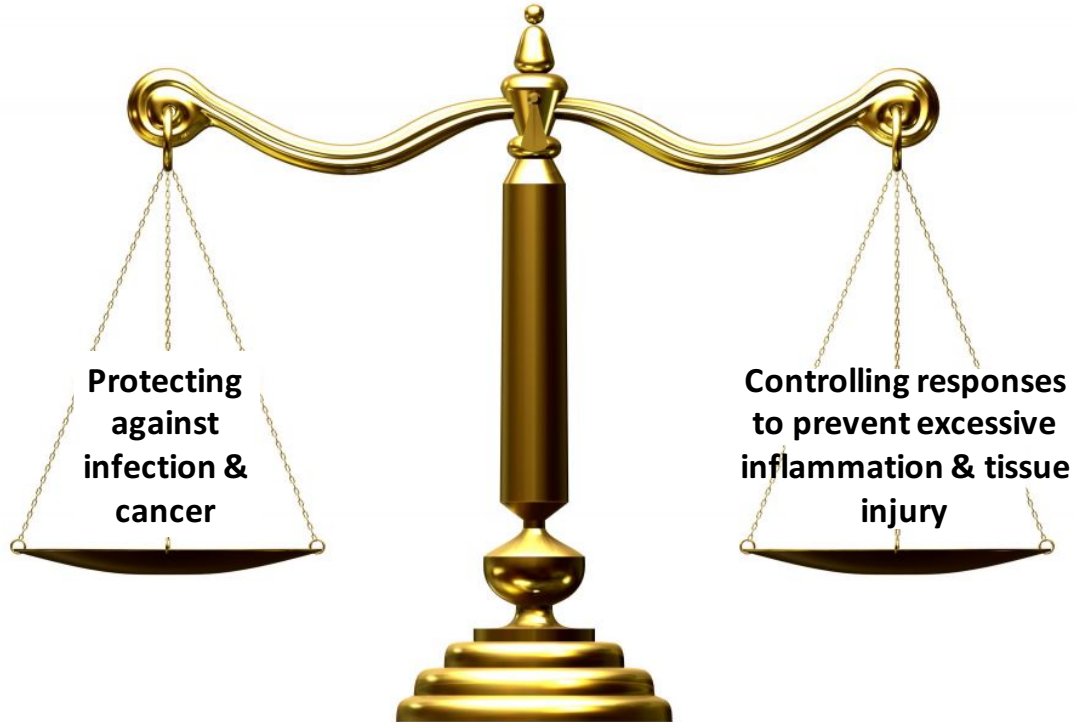
Clinician scientist



Lab

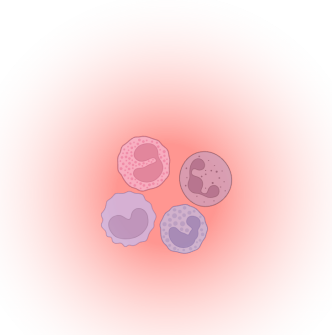


The challenge of the immune system

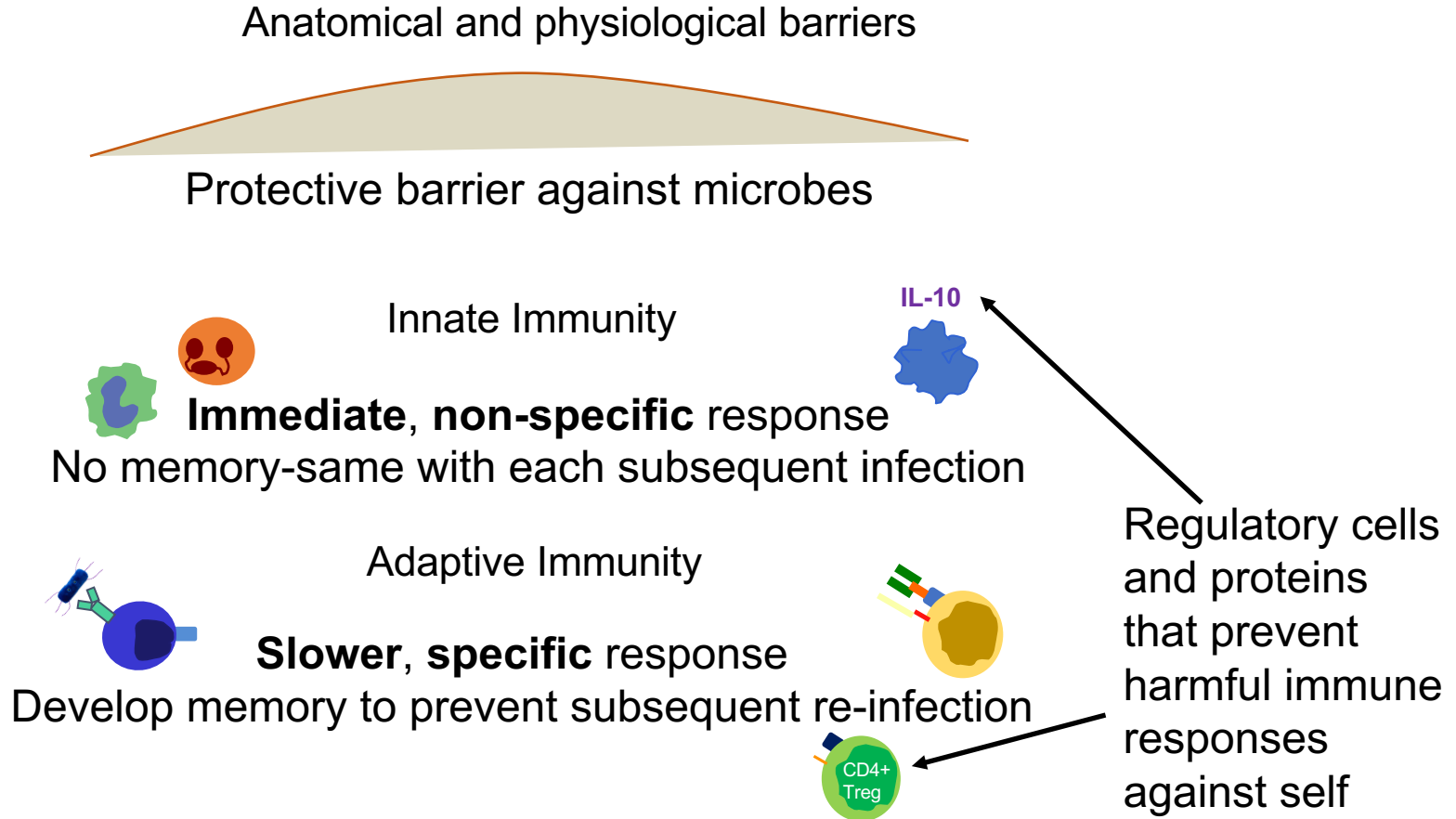


What is inflammation?

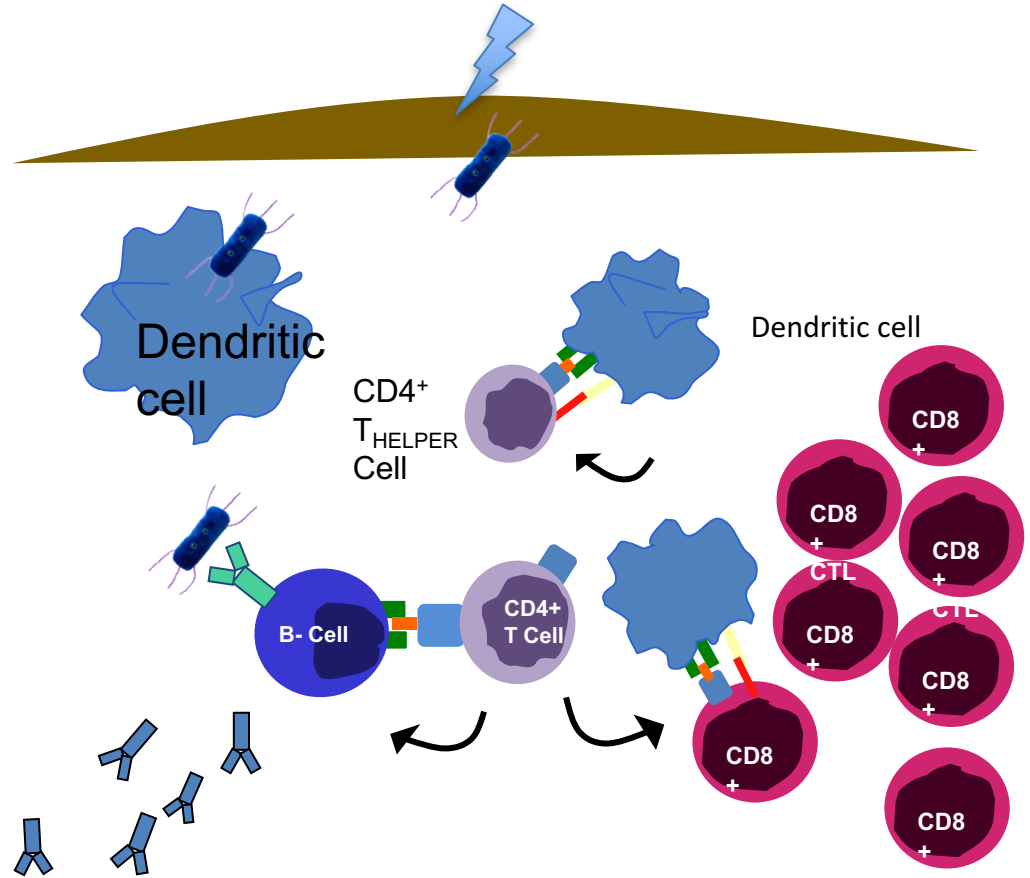
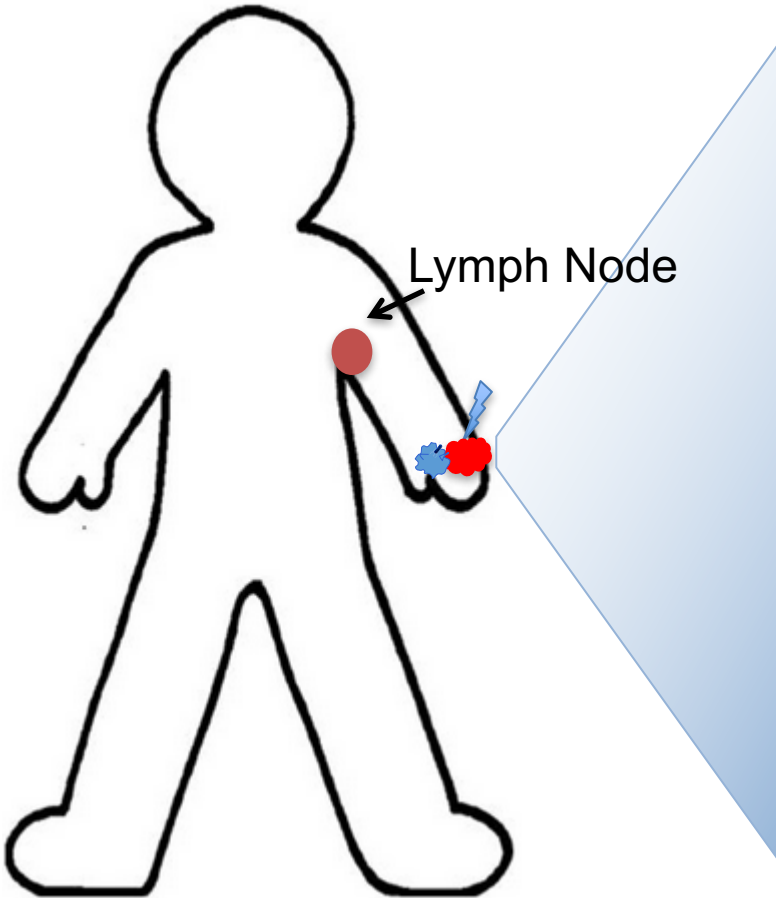
- The body's immune system's response to an irritant
- Ex: germ, cut, etc
- Key signs: redness, heat, swelling, pain



The solution: multi-tiered immune defense



Immune response process



Immune response process

Clearance of infection

Generation of memory T and B cells so that next time a person gets infected with the same microbe, they'll have a faster response

