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HUMAN INBORN ERRORS OF IMMUNITY: AN EXPANDING UNIVERSE

# UNMET NEEDS IN RARE DISEASES, DIFFERING PERSPECTIVES



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X

No, Nothing to disclose

Yes, please specify disclosures

Company / Name	Honoraria / Expense	Consulting / Advisory Board	Funded Research	Royalties / Patent	Stock Options	Ownership / Equity Position	Employee	Other (Please specify)

# UNMET NEEDS OF PATIENTS WITH PID: THE PHYSICIAN PERSPECTIVE

*From your perspective, could you list up  
to 5 unmet needs for PID patients?*

## PREVENT DIAGNOSTIC DELAY

Promote awareness

*Training/Educational programs in PIDs for MDs and Nurses*

Improve screening tools (e.g. NBS for HLH, CGD)

Improve genetic testing availability and turnaround of results

Develop biomarkers

AI-based databases for queries based on sequence and symptoms

# UNMET NEEDS OF PATIENTS WITH PID: THE PHYSICIAN PERSPECTIVE

## ACCESS TO SPECIALIZED CARE

Presence of specialized physicians and referral centers

Access to specialist and recommended/new therapy

Uninterrupted access to Ig therapies

Precision medicine approaches

Improved/tailored strategies for prevention and management  
of infections and co-morbidities

Nursing participation to management

# UNMET NEEDS OF PATIENTS WITH PID: THE PHYSICIAN PERSPECTIVE

## PSYCHOLOGICAL SUPPORT

Peer groups

Mental health services

Counseling,

Patient education programs

## TRANSITION OF CARE

# UNMET NEEDS OF PATIENTS WITH PID: THE PHYSICIAN PERSPECTIVE

## SPECIFIC THERAPEUTIC NEEDS

Methods to predict and monitor outcome severity

Improved/tailored strategies for prevention and management of  
infections and co-morbidities

Anti-inflammatory treatments devoid of immunosuppressive effects

Methods to accelerate healing and regeneration after anti-  
inflammatory treatment

Methods to cure GOF diseases with gene therapy

# UNMET NEEDS OF PATIENTS WITH PID: THE PHYSICIAN PERSPECTIVE

## EXTRA MEDICAL NEEDS

Help with socio-economic burden

Lack of political support in resource-limited  
situations/environments

Increase opportunity to report from resource-limited  
situations/environments

Political action to advocate for lower and capped costs  
of therapeutics



# UNMET NEEDS OF PATIENTS WITH PID: THE PHYSICIAN PERSPECTIVE

## RESEARCH-RELATED NEEDS

Worldwide registries (symptoms, new findings, treatment efficacy)

Right to be included and research studies

Streamlining of regulatory requirements on research studies

Understanding of the pathophysiology of the disease



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