



National Human
Genome Research
Institute



National
Institutes of
Health



U.S. Department
of Health and
Human Services

Research Directions in Genetically Mediated SJS/TEN

March 3-4, 2015

*National Institutes of Health Campus
Bethesda, Maryland*



Teri Manolio, M.D., Ph.D.
Director, Division of Genomic Medicine

Joint Effort and Collaborative Funding

- Food and Drug Administration (FDA)
- National Center for Advancing Translational Sciences (NCATS)
- National Institute of Allergy and Infectious Diseases (NIAID)
- National Institute of Arthritis, Musculoskeletal and Skin Diseases (NIAMS)
- National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK)
- National Institute of Neurologic Disorders and Stroke (NINDS)
- National Human Genome Research Institute (NHGRI)

Research Directions in Genetically-Mediated SJS/TEN

- How did we get here?
- What are our goals?
- Where do we go from here?
- How do we get there?
- Where are the bathrooms....



National Academy of Sciences Bldg
2101 Constitution Avenue, NW
Washington, D.C.

NHGRI's Genomic Medicine Meetings

Genomic Medicine Centers Meeting VI: *Global Leaders in Genomic Medicine*

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On January 8-9, 2014, the National Human Genome Research Institute (NHGRI), sponsored its sixth Genomic Medicine Centers meeting - *Genomic Medicine Centers Meeting VI: Global Leaders in Genomic Medicine* - at the National Academy of Sciences Building in Washington, D.C. Geoffrey Ginsburg, M.D., Ph.D., Duke University, and Teri Manolio, M.D., Ph.D., NHGRI, co-chaired the meeting.

The goals of the meeting were to:

- Identify areas of active translational and implementation research, potential common strategies, and opportunities for collaborative efforts.
- Identify common barriers to implementation of genomics in healthcare and a policy agenda relevant to advances in the field.
- Identify nations with unique capabilities (such as national healthcare systems) that may allow rapid implementation and measures of key outcomes.
- Discuss opportunities (such as national healthcare system) that may allow rapid implementation and measures of key outcomes.

[Genomic Medicine 6 Executive Summary](#) 

[Genomic Medicine 6 Full Meeting Minutes](#) 

View meeting videos and slides from: [January 8](#) [January 9](#)

 [Video Playlist](#)

Wednesday, January 8, 2014

<http://www.genome.gov/27549225>

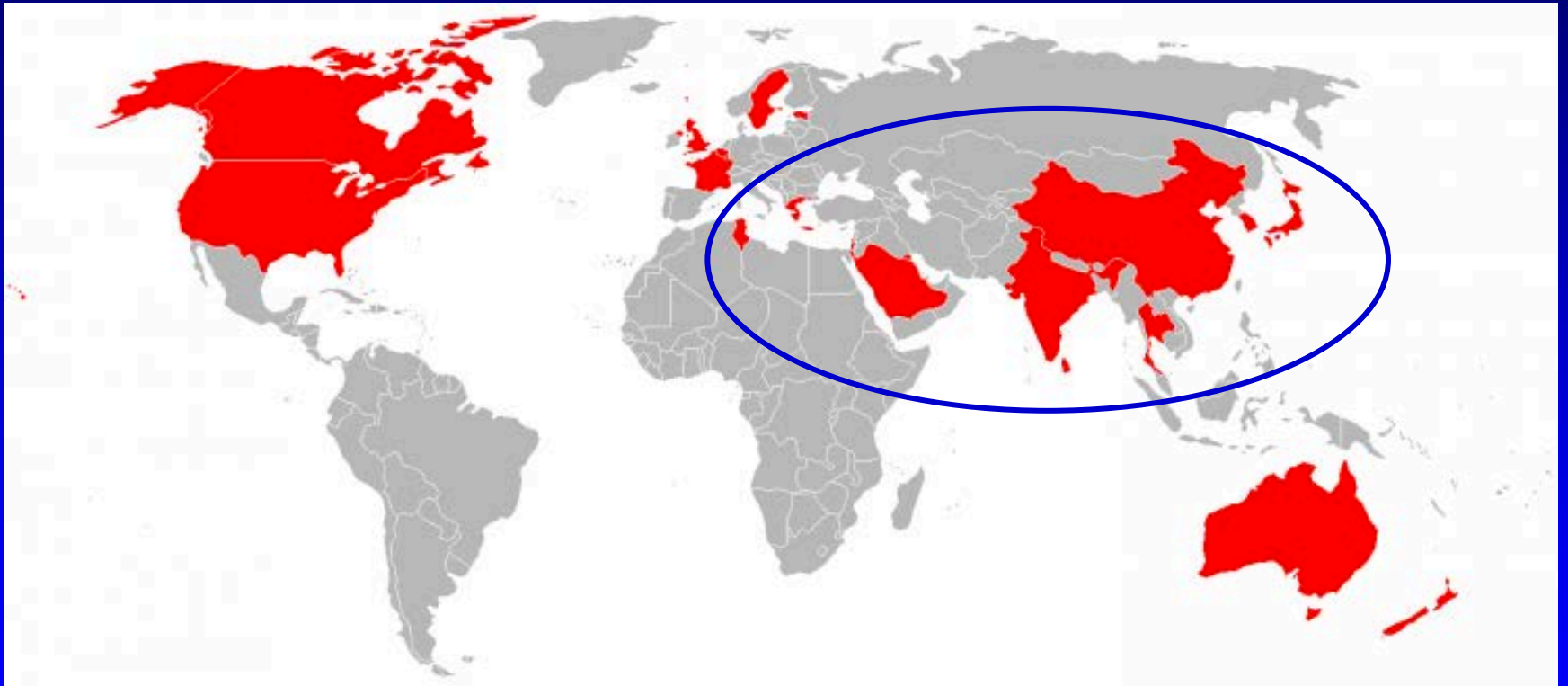
50 International Genomic Medicine Leaders



Global Leaders in Genomic Medicine
Washington, DC, USA
January 8, 2014

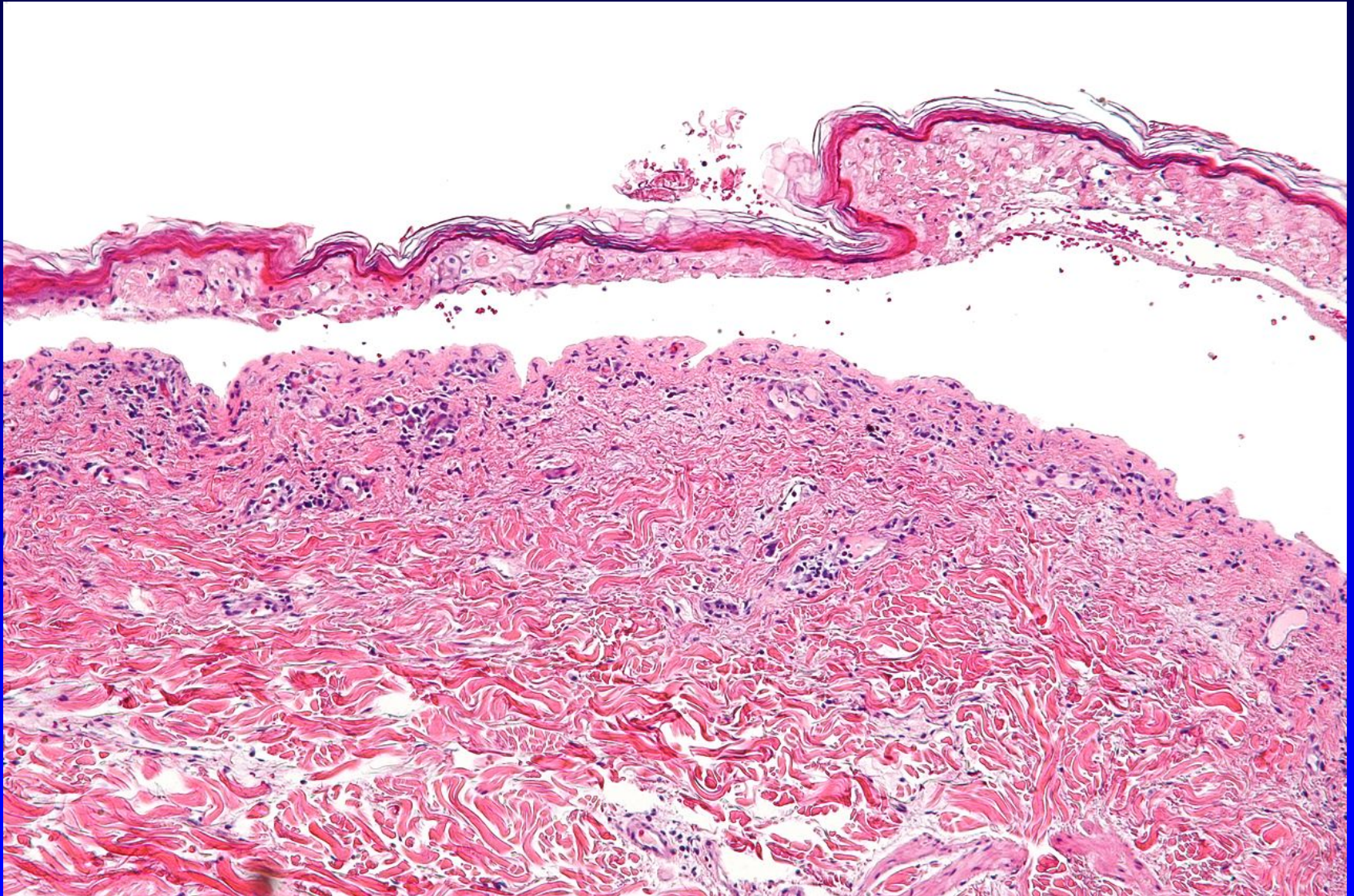
40 US Genomic Leaders and NHGRI Staff

50 International Genomic Medicine Leaders 25 Countries

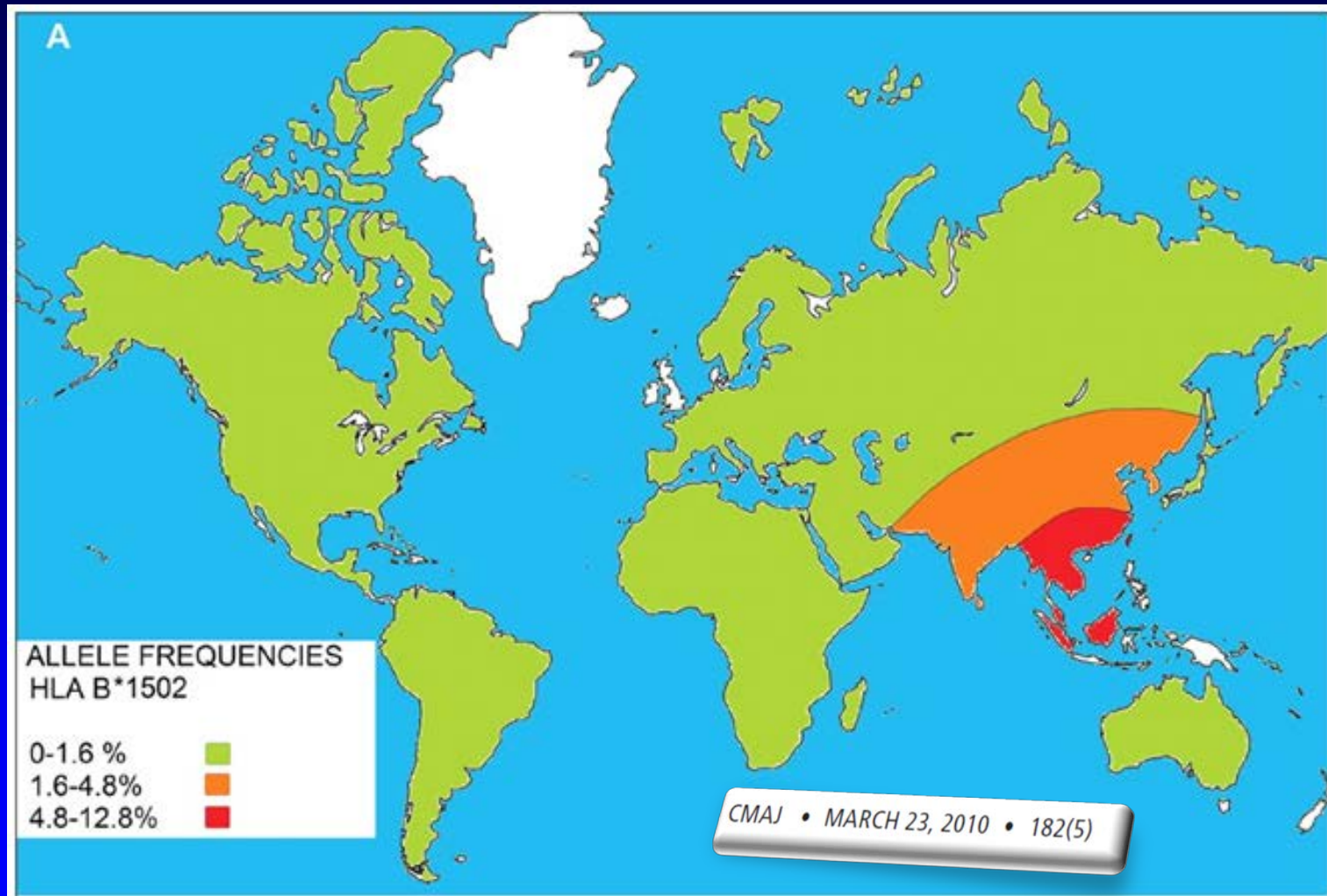


Courtesy, G Ginsburg, Duke U

Stevens Johnson Syndrome/Toxic Epidermal Necrolysis



Carbamazepine and SJS/TEN: Allele Frequency of HLA-B*15:02



Courtesy W Chantratita, Ramathibodi Hospital



เภสัชพันธุศาสตร์และการรักษาเฉพาะบุคคล
คณะแพทยศาสตร์ โรงพยาบาลรามธิบดี

[Redacted Name]

ผลการตรวจ: HLA-B Gene : HLA-B*15:02/15:25

วันที่ตรวจ: 8 มกราคม 2557

การแปลผลทางเภสัชพันธุศาสตร์:

ตรงกับตัวบ่งชี้ต่อการแพ้ยา Carbamazepine ตามฐานข้อมูลในปัจจุบัน

Name & Family Name

Outcome of the PGX assay

8 Jan 2014

PGx Interpretation

High Risk of SJS/TEN from Carbamazepine, according to update information

Suggestion: According to update information, this person has HLA-B*1502 which has a high risk to develop a severe skin disorder (SJS/TEN), if he takes carbamazepine or drug structurally similar.

Need more information: please contact our PGx laboratory. Tel 02-200-4330-3...

Courtesy W Chantratita



Pharmacogenomics and Personalized Medicine
Faculty of Medicine Ramathibodi Hospital

ข้อเสนอแนะ ผลการตรวจยีน HLA-B พบความสัมพันธ์กับตัวบ่งชี้ต่อการแพ้ยาตามฐานข้อมูลในปัจจุบันคือ HLA-B*15:02 ซึ่งมีความสัมพันธ์กับการเกิดอาการแพ้ยาทางผิวหนังชนิดรุนแรง (Stevens-Johnson syndrome และ Toxic epidermal necrolysis) ดังนั้นไม่ควรใช้ยา Carbamazepine หรือยาที่มีสูตรโครงสร้างใกล้เคียงในผู้ป่วยรายนี้

ต้องการข้อมูลเพิ่มเติม ติดต่อ: หน่วยเภสัชพันธุศาสตร์และการรักษาเฉพาะบุคคล
โทรศัพท์ 02-200-4330-3 หรือ 02-201-1380, 02-201-1390

Signature of molecular clinical pharmacist.

ภก.ดร.ชลภัทร สุขเกษม

Workshop on Research Directions - Objectives

1. Review current state of knowledge of surveillance, pathogenesis, and treatment
2. Examine role of genomics and PGx in etiology, treatment, and eradication of preventable cases
3. Identify gaps, unmet needs, and priorities for future research to eliminate SJS/TEN globally

Planning Group

Mark Avigan, FDA
Ricardo Cibotti, NIAMS
Robert Davis, U Tenn
Josh Denny, Vanderbilt

Carolyn Hutter, NHGRI
Lois La Grenade, FDA
Neil Shear, U Toronto
Lisa Wheatley, NIAID

Agenda Review

Tuesday, March 3rd 8:30 AM - 5:45 PM

8:30 Session 1: State of the Science

11:30 Session 2: International Experience Part 1

Includes working lunch

1:30 Session 3: Challenges in Case Finding and Surveillance

4:00 Session 4: Working Groups

Working group and room assignments are in your packet

Agenda Review, Day 2

Wednesday, March 4th 8:30 AM - 3:00 PM

8:30 Session 5: International Experience Part 2

10:30 Session 6: Special Topics

Includes working lunch

12:45 Session 7: Report of Working Groups

2:15 Session 8: Wrap-up and Next Steps

3PM Adjourn, return to hotel or to airports

Video Livestream

- The workshop is being broadcast live, and will be video archived
- A link to the video is available at: <https://www.genome.gov/27560487>
- Please use microphones

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Advancing human health through genomics research

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Research Directions in Genetically-Mediated Stevens-Johnson Syndrome/Toxic Epidermal Necrolysis

March 3 - 4, 2015

Building 31 Conference Center (6th floor of 31C)
NIH Main Campus
Bethesda, Md.

On March 3-4, 2014, the National Human Genome Research Institute (NHGRI) will sponsor a workshop - *Research Directions in Genetically-Mediated Stevens-Johnson Syndrome/Toxic Epidermal Necrolysis* - at Building 31 on NIH Main Campus.

The objectives of the workshop are to:

1. Review current state of knowledge of surveillance, pathogenesis, and treatment of Stevens-Johnson Syndrome/Toxic Epidermal Necrolysis (SJS/TEN)
2. Examine role of genomics and pharmacogenomics in etiology, treatment, and eradication of preventable causes of drug-induced SJS/TEN
3. Identify gaps, unmet needs, and priorities for future research to eliminate genetically mediated SJS/TEN globally

Watch it live here, March 3rd at 8:30 a.m. Eastern

See presenters on: [March 3](#) [March 4](#)

Tuesday, March 3, 2015

Location: NIH Building 31 Conference Center 6C Room 10

Time	Topic	Moderator/Presenter
8:00 a.m.	Registration	
Session 1: State of the Science Moderator: Ricardo Cibotti		
8:30 a.m.	Welcome, Introduction and Goals	Teri Manolio
9:00 a.m.	Clinical syndromes, epidemiology, genomics, diagnosis and treatment	Neil Shear
9:30 a.m.	Basic science of pathogenesis, functional genomics and mechanisms	Wen-Hung Chung
10:00 a.m.	Defining and prioritizing unmet research needs for a rare but deadly disease	Elizabeth Phillips
10:30 a.m.	Discussion	All
11:00 a.m.	Break	

IN CONGRESS, JULY 4, 1776.

The unanimous Declaration of the thirteen united States of America.

- We apologize that government regulations prohibit us from paying for meals
- There is a cafeteria on the first floor
- If you ordered lunch on line, it will be delivered to the meeting room each day
- We hope you can join us for dinner tonight at Bistro Provence!
 - Sign up at registration table by end of 1st break
 - Map in your packet



Logistical Support

- Registration Desk
 - Jennifer Adona and Joshua Shapiro from Capital Consulting Corporation
 - Assistance with logistical questions
 - Assistance logging onto NIH guest network
 - Taxi arrangements
- Shuttle
 - Shuttle to hotel from B2 level at 6:00 PM
 - Shuttle will pick up at hotel at 7:15 AM

Thank You!

* On both the Planning Committee and the Working Group

Objectives of GMVI: Global Leaders in Genomic Medicine

- Identify areas of active translation and implementation
- Prioritize common barriers to implementation in healthcare
- Frame a policy agenda to advance the field
- Highlight nations with unique capabilities
- Discuss opportunities for international collaborations