



SCHOOL OF MEDICINE

Program in Immunology

Department of Medicine

Division of Clinical Immunology and Rheumatology

Genomics and Epigenetics of Common Variable Immune Deficiency & REcurrent SinoPulmonary Infections (CVID & RESPI)

Harry W Schroeder Jr MD PhD, PI
Professor of Medicine, Microbiology and Genetics
Director, UAB Program in Immunology
University of Alabama at Birmingham

Elizabeth E Brown PhD MPH, Co-PI
Associate Professor of Public Health
University of Alabama at Birmingham

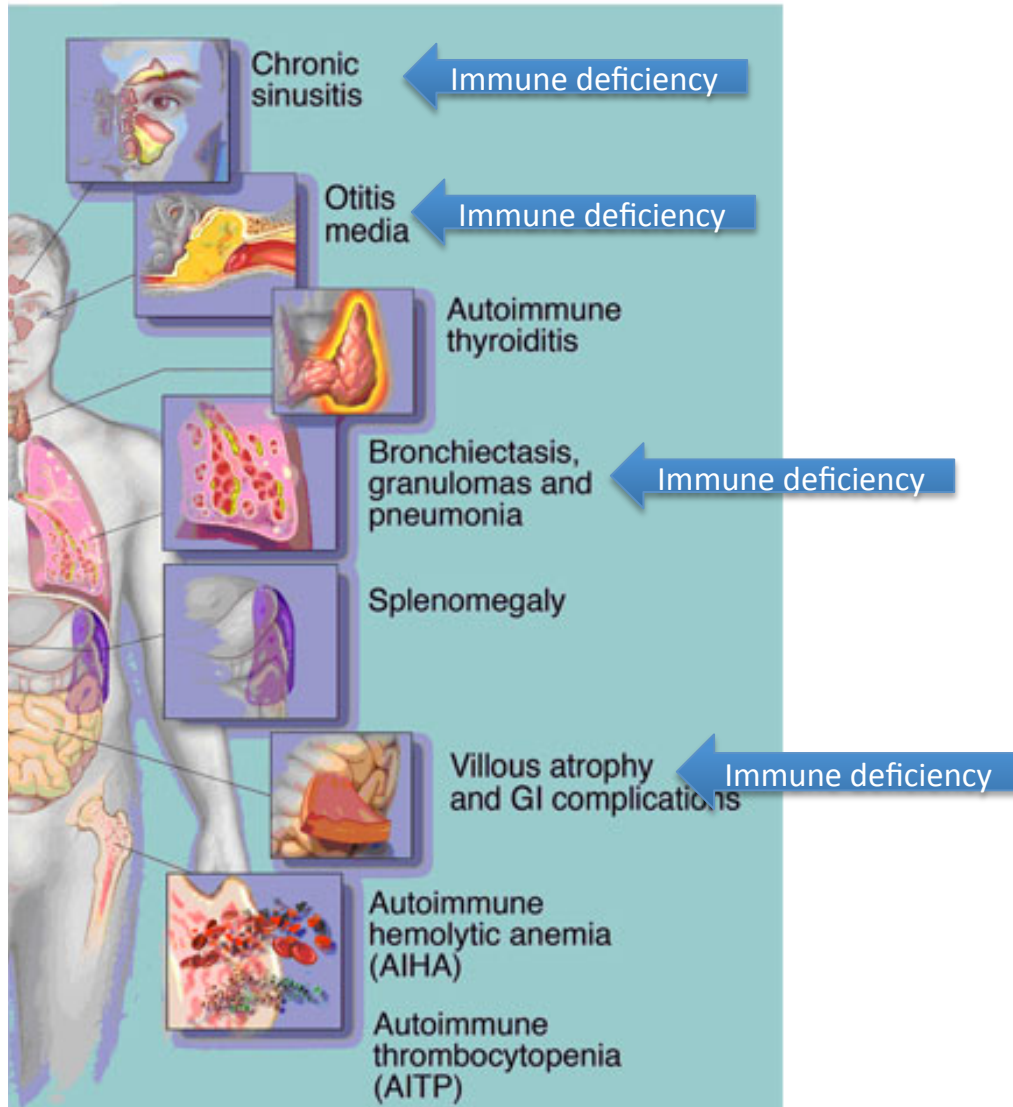


A Typical History

- High School - Catches every cold
- College – Repeated Sinusitis & Bronchitis
 - First Sinus Surgery
- First Job – First Pneumonia
- Depression, Chronic Fatigue, Always Sick
- Develops hypo- or hyperthyroidism
- Pneumonia #3, Sinus Surgery #5, or Cushingoid due to Steroid Therapy

CVID is a Multi Organ and System Disease

Clinical anomalies

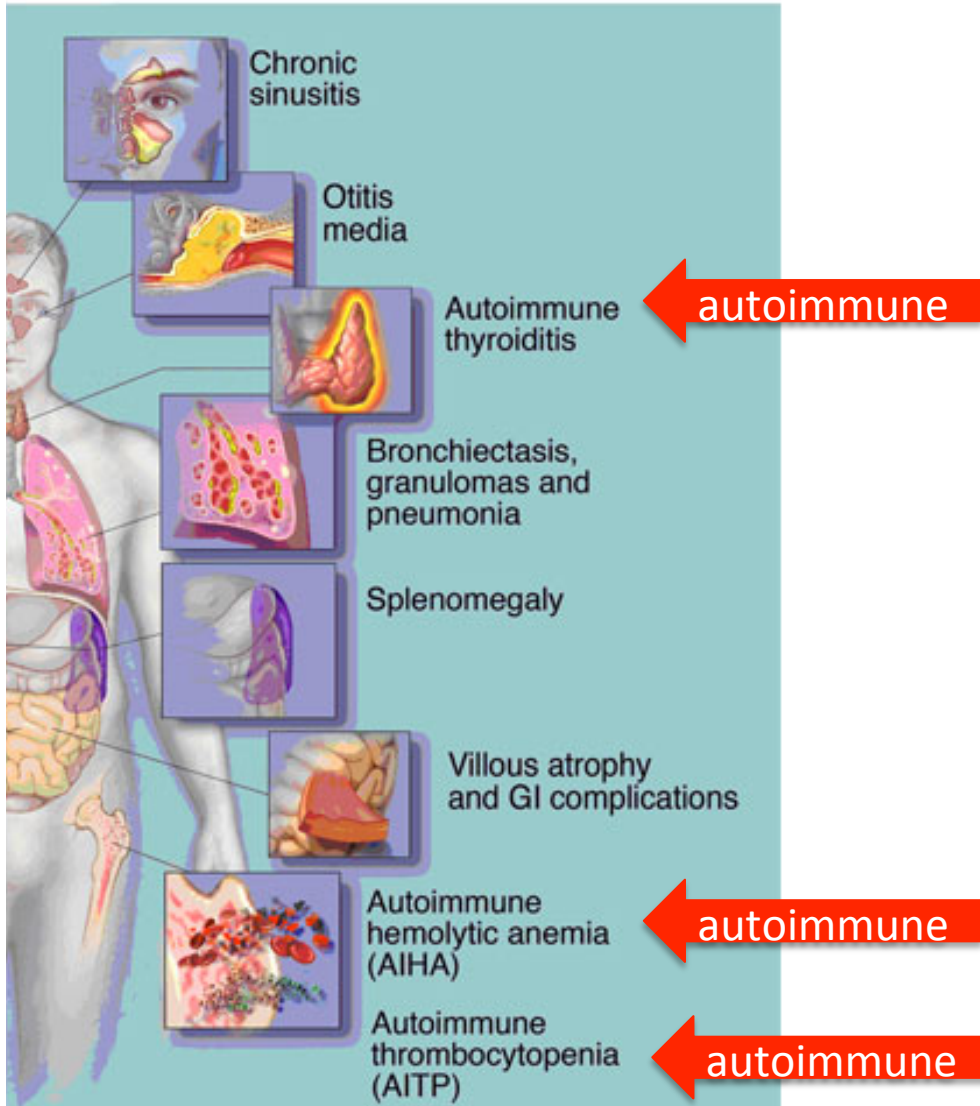


Immune deficiency in CVID

- Decreased B cells
 - Blunted /absent antibody titers
 - Reduced Ig Levels
- ↓
- 75% of patients develop chronic pulmonary structural complications
 - Recurrent infection
 - Tissue damage
 - GI complications
 - Increase gastric cancer and
 - Non Hodgkin lymphoma (1.5-7%)

CVID is a Multi Organ and System Disease

Clinical anomalies

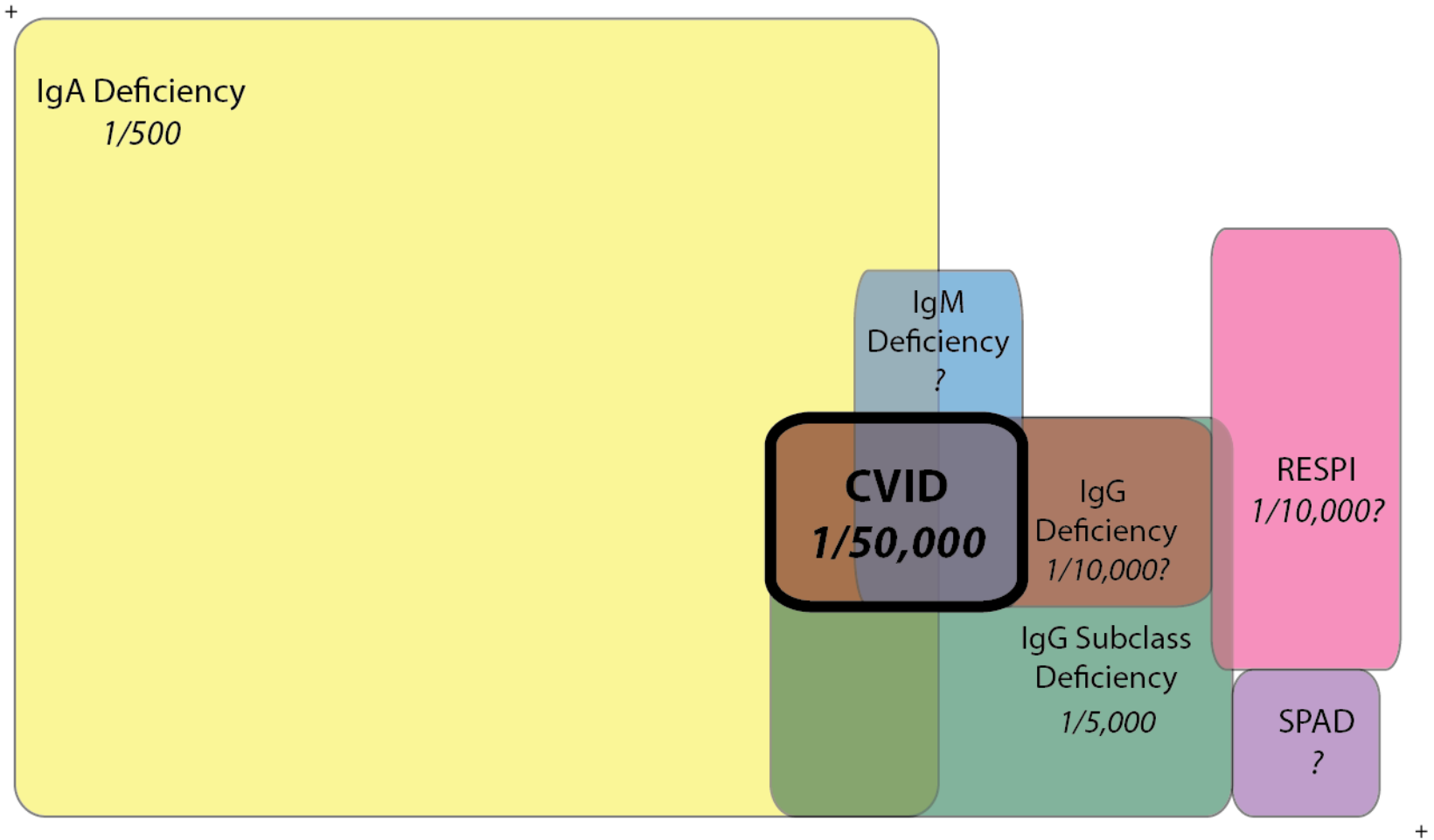


Autoimmune in CVID

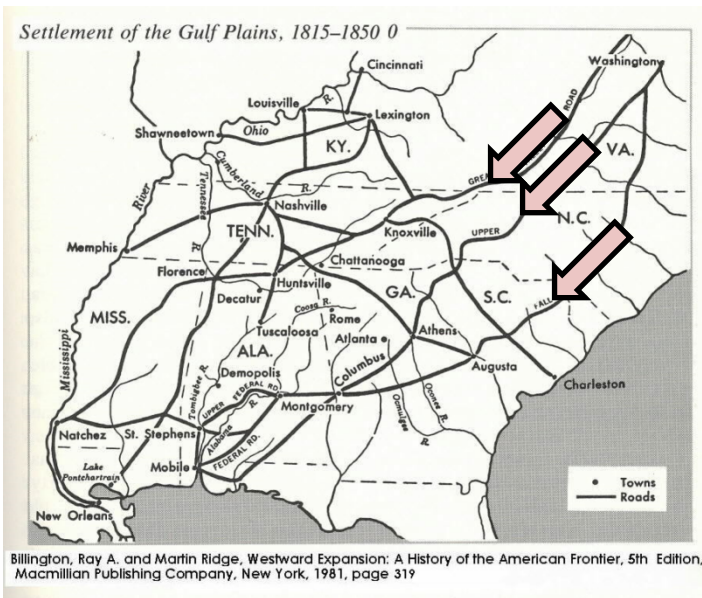
- 20% of CVID patients have some form of autoimmunity
- Cytopenias are more common presenting disorder in children than adults

- Pernicious anemia
- Autoimmune neutropenia
- Graves disease
- Hypothyroidism
- Evans syndrom
- SLE
- RA

Spectrum of Antibody Deficiency



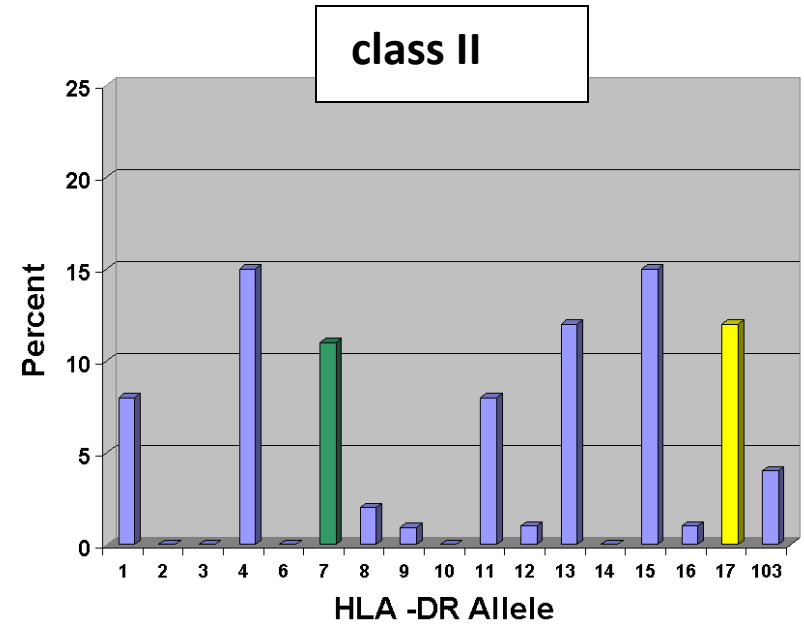
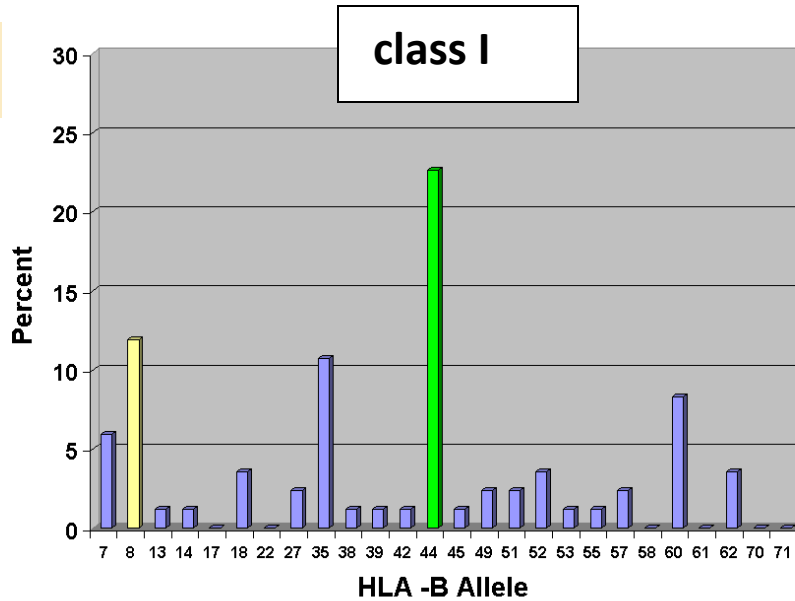
Demographics - Greater Alabama



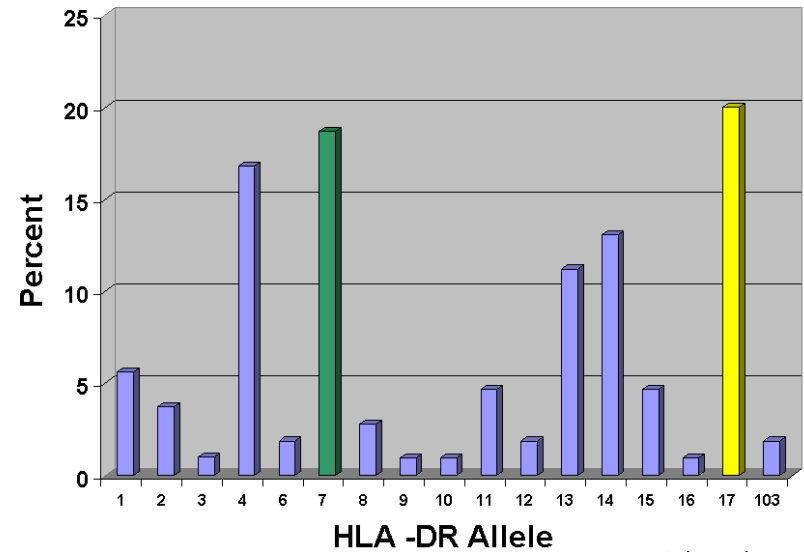
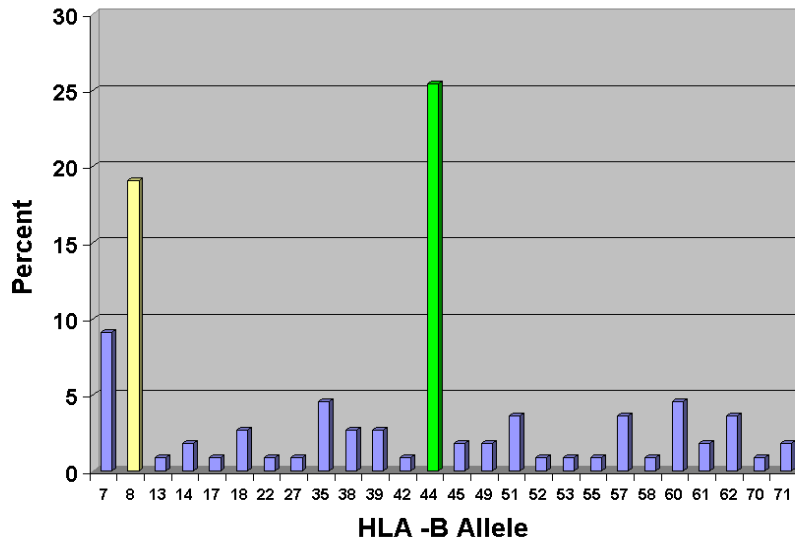
- 4,000,000 people in the catchment area
- Most European-Americans a product of two waves of emigration
 - (Scottish Borders to Appalachian Piedmont, then to Alabama)
- 3rd most stable population in the US

Pathogenesis of CVID/RESPI

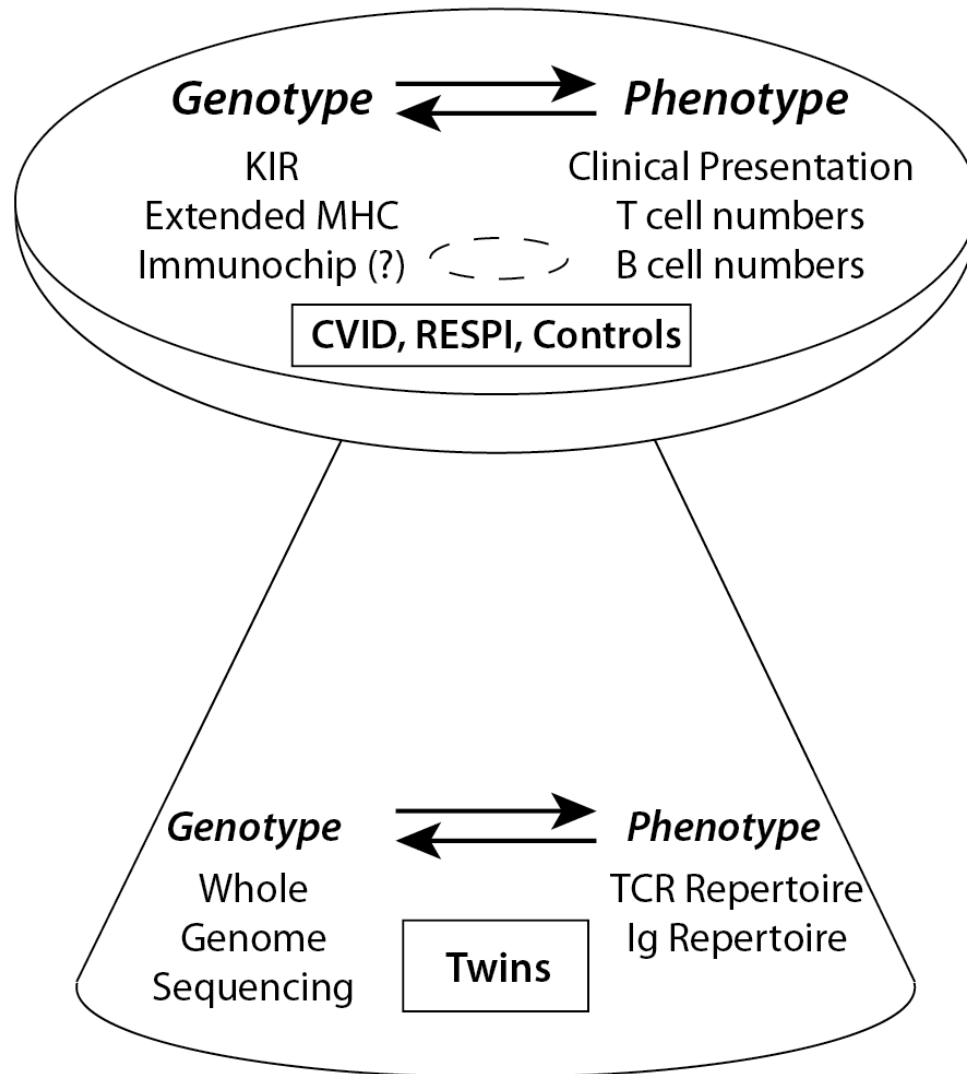
RESPI



CVID



A Wide and Deep Analysis



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