



# Rishi Vansh

All India Institute of  
Medical Science  
Rishikesh AIIMS R

Department Of

Pediatric Genetic -division

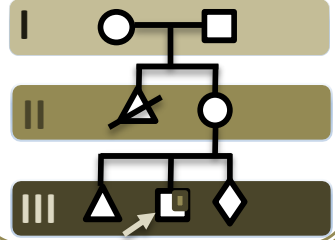
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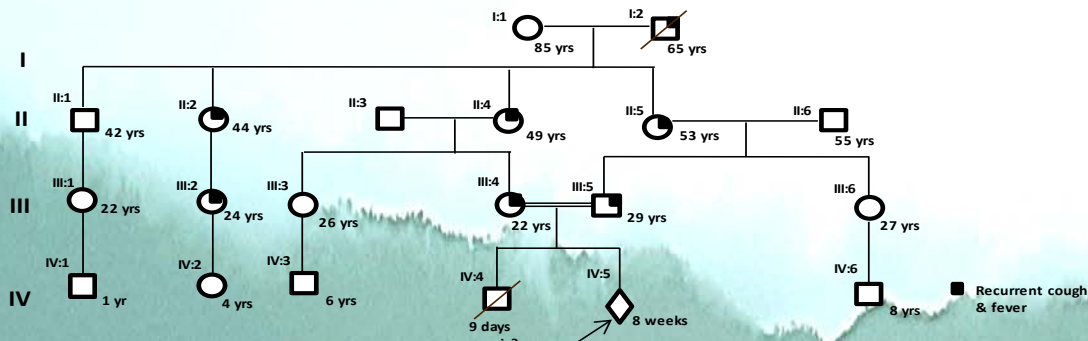


### From the desk of Editor

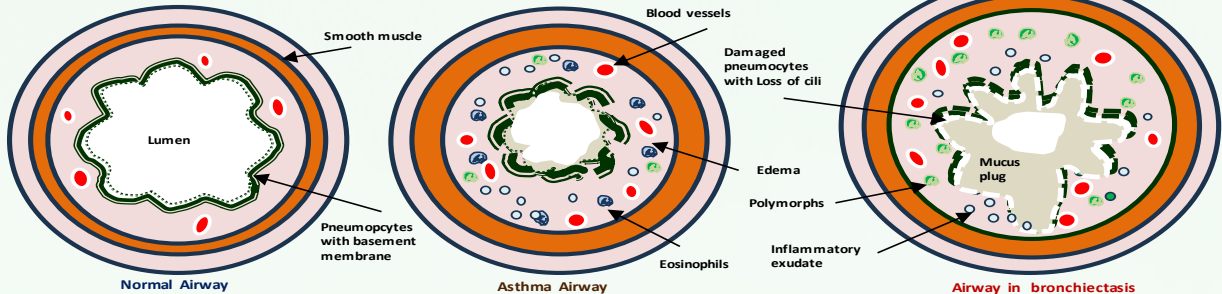
The genetic division of the Pediatric Department is publishing a monthly newsletter for faculty and residents. The newsletter is related to genealogical parlance and is a deliberate attempt to enhance awareness of genetic disorders with recent updates.

## Pulmogenetics-(V)

### Human Genetics/ Genetics with "Bronchiectasis" – Part-I



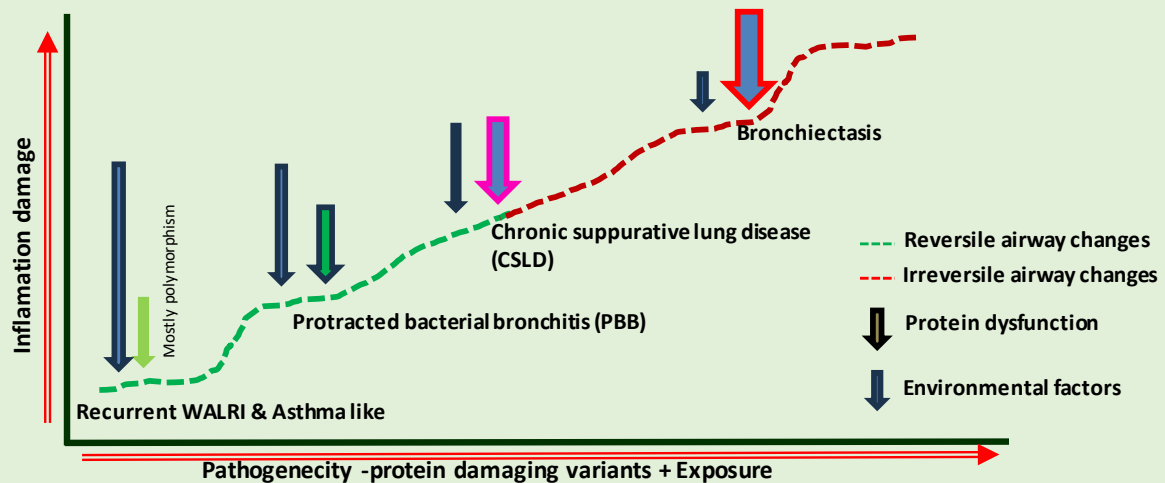
### Basic pathogenicity from reactive airway disease to Bronchiectasis



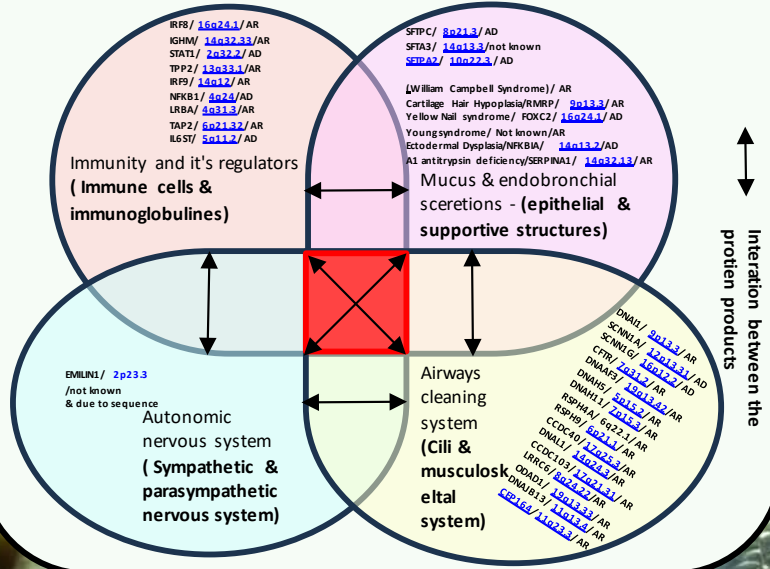
### Insight:

1. What is bronchiectasis, and what is its clinical presentation?
2. How can the same gene dysfunctions lead to asthma to bronchiectasis?
3. What is the endobronchial immune reactivity spectrum disorders?
4. What is the possible role of lifestyle modification on recurrent airway disease?
5. How would you antenatal counselling of the family for case IV:5?

## Endobronchial immune reactivity: reversible changes (Asthma) to irreversible damage (Bronchiectasis)



### Genes related to Normal Endobronchial functioning



### A child with recurrent cough

Clinical syndrome	Clinical diagnosis	Lab	Radiology
Asthma	Recurrent & Reversible airway limitation	≥200 mL or ≥12% changes from the baseline FEV1 with bronchodilator.	Normal
Protracted bacterial bronchitis (PBB)	Antibiotics responded Wet Cough 2-4 weeks	Evidence of bacterial infection	Structural defects may be present
Chronic suppurative lung disease (CSLD)	Chronic wet cough >4 week & clinical features of chronic hypoxia (growth, PAH, clubbing)	Evidence of chronic suppurative infection - defective endobronchial clearance system	Broncho-vascular thickening and the inner airway and the outer vessel diameter ratio < 0.80 in CT
Bronchiectasis	Same as CSLD	Same as CSLD	Ratio > 0.80 in CT

The complex interaction of proteins function responsible for above-overlapping phenotypes with different grading of severity- complex "PheneGene Graphics" also displayed in OMIM helpful to understand this interaction at genes level in both linear (depth of 4 nodes) & radial format

**Counseling the family for case IV:5-** Counsel for next generation based molecular testing for "panel test of genes related to chronic cough" and "polymorphic marker screening" with its limitation, even for a common multifactorial trait with a positive family history of pathological phenotypes. Theoretically, there is a strong possibility of homozygosity for relevant polymorphic markers or mild variant mutants in the fetus with severe clinical disease.

### Thought Riveting:

- What is the weightage of emotional stress in Pediatric Asthma as a precipitating factor?
- Does non-antibiotic derivatives of azithromycin work as modifiers at the transcription level?
- How does multigene "the threshold model" responsible for "non-CF-chronic cough spectrum disorders" in children?
- How much will be ethically acceptable & clinically relevant to do genomic analysis for polymorphic markers in a child with chronic cough?
- Can the lifestyle modification (healthy diet, adequate physical activity, and positive mental health change) outdate the use of allopathic medicines in a child with chronic cough with reversible airway disease?

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