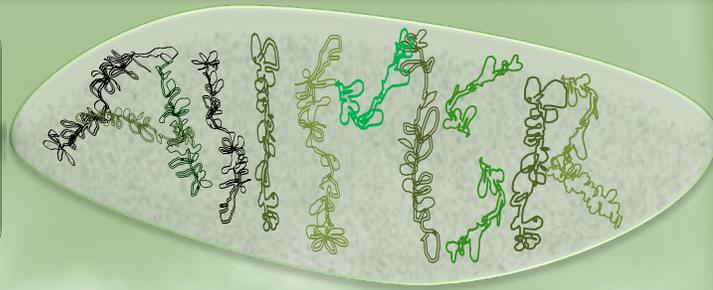




# Rishi Vansh

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Reviewer: Dr. Raksha Ranjan

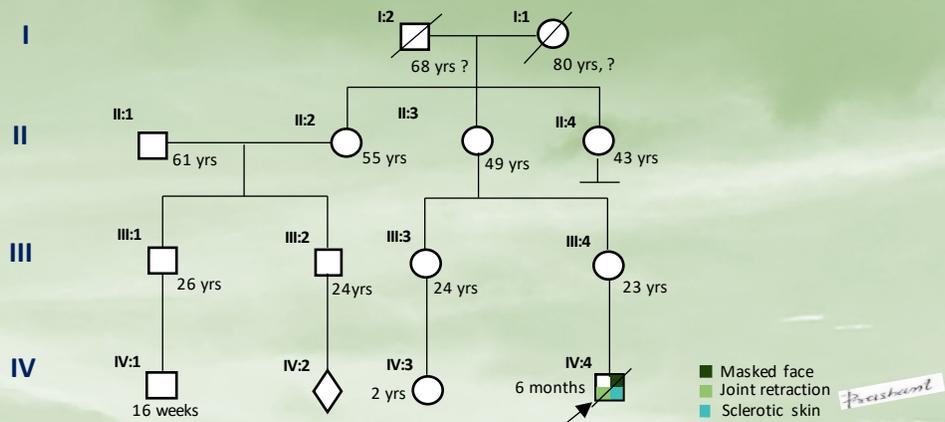
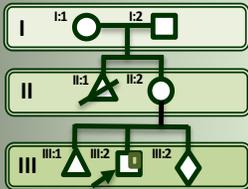
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## Progeroid syndromes (PS)-VII / LAMIN A/C related disorders / Restrictive dermopathy 2

From the desk of Editor

The genetic division of the Pediatric Department publishes a monthly newsletter for all Medical Professionals. The newsletter is related to genealogical parlance and is a deliberate attempt to enhance awareness of genetic disorders with recent updates.



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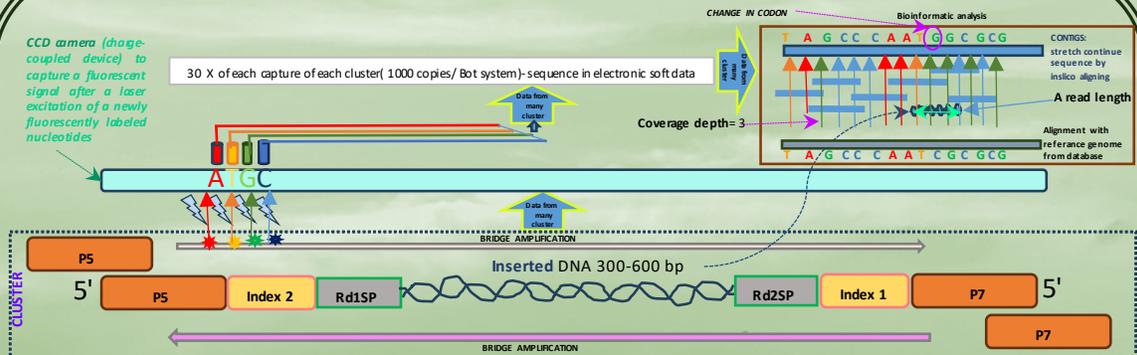
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### Basic aspect of Sequencing by Synthesis (SBS)



Rd1SP/ Rd2SP- Binding site for sequence primers  
P5/P7- Complementary probes to oligos of illumina NGS platform  
Index 1/2- Barcode or Tags, 8-12 bp, unique for particular sample, make possible multiplexing (run many samples together)  
Cluster- A clonally amplified specific sequence templet which makes paired-end read (≈2 K) through repetitive amplification  
Coverage depth - number of reads for a reference codon  
Read length- number of base pairs(bp) that are sequenced in a single read

### Insight:

1. How would you do periconceptual counselling of Case III: 4?
2. What is the basic principle of Single-Read Sequencing (SRS)?
3. What are the differences between Type 1 and Type 2 restrictive dermopathy?
4. How would you approach a case with arthrogyriposis?
5. What is the Peptidase M48 family?

