MAYO CLINIC

CLINICAL VARIANT INTERPRETATION LAB

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Introduction to Computational Genomics
June 24th, 2024

Case 2: Group Walkthrough #1

In breakout groups, walk through your variants and apply criteria you think are appropriate based on publically available databases.

Case #2

Patient phenotype:
- Retinitis pigmentosa

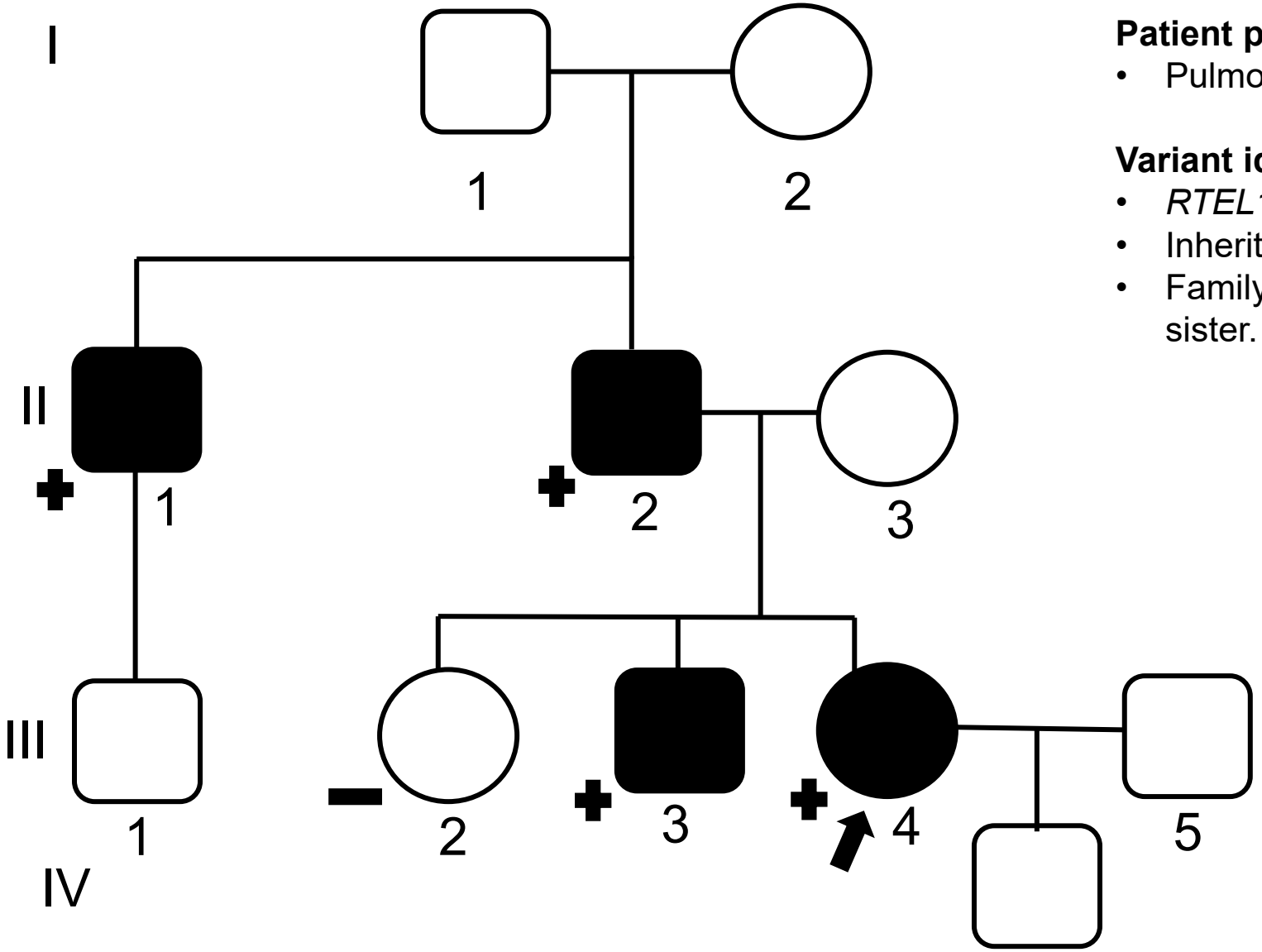
Variant identified via trio exome:
- *RPGR:*c.905G>A, Cys302Tyr
- Transcript: NM_001034853.2
- Inheritance: Unknown, but variant is on the X-chromosome

Criteria being considered	Strength being applied	Evidence	Points

Case 3: Group Walkthrough #2

In breakout groups, walk through your variants and apply criteria you think are appropriate based on publically available databases.

Variant #3



Patient phenotype:

- Pulmonary fibrosis, shorted telomeres

Variant identified via trio genome:

- *RTEL1:*c.101A>G, Q34R
- Inheritance: Paternal
- Family history: 1 affected brother, 1 unaffected sister. Father and paternal uncle are affected.

Variant #3

Patient phenotype:
- Pulmonary fibrosis, shorted telomeres

Variant identified via trio genome:
- *RTEL1:*c.101A>G, Q34R
- Inheritance: Paternal
- Family history: 1 affected brother, 1 unaffected sister. Father and paternal uncle are affected.

Criteria being considered	Strength being applied	Evidence	Points

Case 4: Group Walkthrough #3

In breakout groups, walk through your variants and apply criteria you think are appropriate based on publically available databases.

Case #4

Patient phenotype:

- Developmental regression, ataxia, seizures, cerebellar atrophy, nystagmus, Dandy-Walker malformation

Variant identified via trio genome:

- *In trans* variants:
  - *PMPCA*:c.1204C>T, R402*
  - *PMPCA*:c.667C>T, R223C

Criteria being considered	Strength being applied	Evidence	Points
Variant 1 R402*			
Variant 2 R223C			

QUESTIONS & ANSWERS