



Brigham and Women's Hospital



Harvard Medical School

Molecular Genetic Research Unit

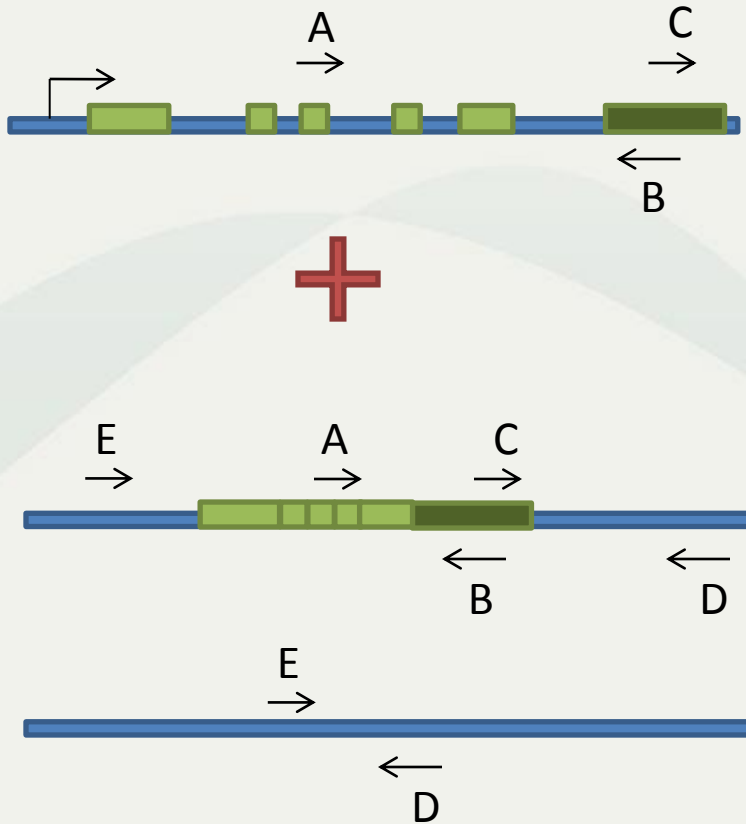
Validation of Polymorphic Processed Pseudogenes

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Primer Design

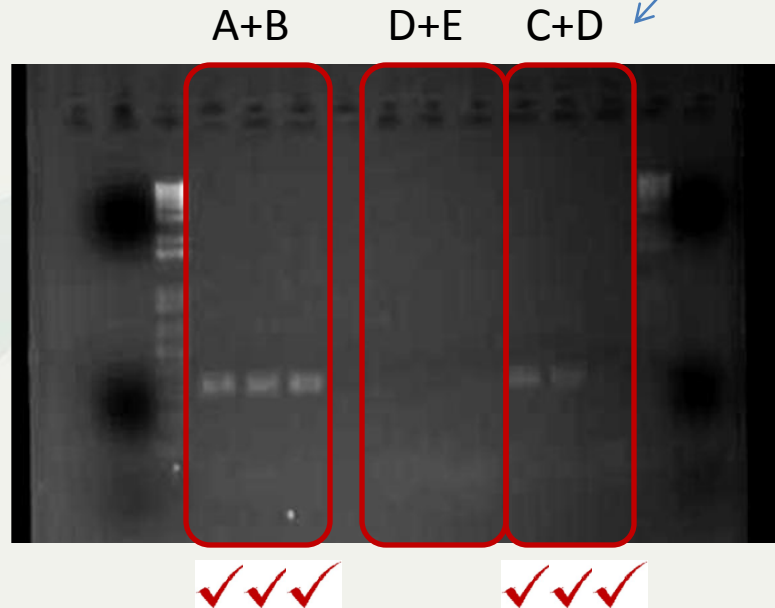


All pgene insertions predicted to be on minus strand.

- A + B pgene exists
- E + D no pgene at predicted site (for ≥ 1 chr)
- C + D pgene at predicted site (for ≥ 1 chr)

CBX3

Predicted size = ~340 bp



- A + B pgene exists
- E + D no pgene at predicted site (for ≥1 chr)
- C + D pgene at predicted site (for ≥1 chr)

All 3 in the CEU trio have the pgene in the correct location.

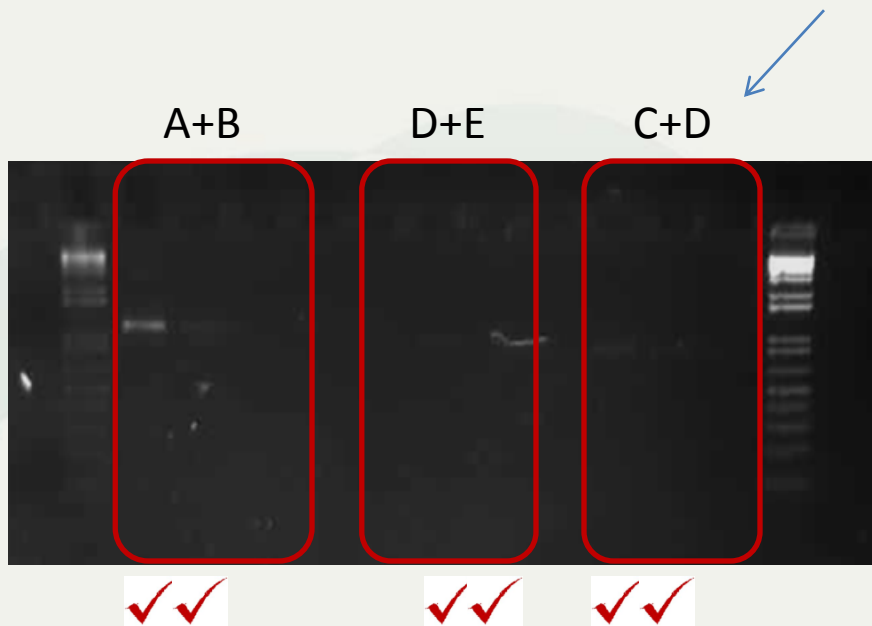
Parent gene id	Name	NA12878	NA12891	NA12892
ENSG00000122565	CBX3	Yes	Yes	Yes
ENSG00000139372	TDG	Yes	No	No
ENSG00000165480	SKA3	Yes	Yes	No





TDG

Predicted size = ~840 bp



A + B pgene exists
 E + D no pgene at predicted site (for ≥ 1 chr)
 C + D pgene at predicted site (for ≥ 1 chr)

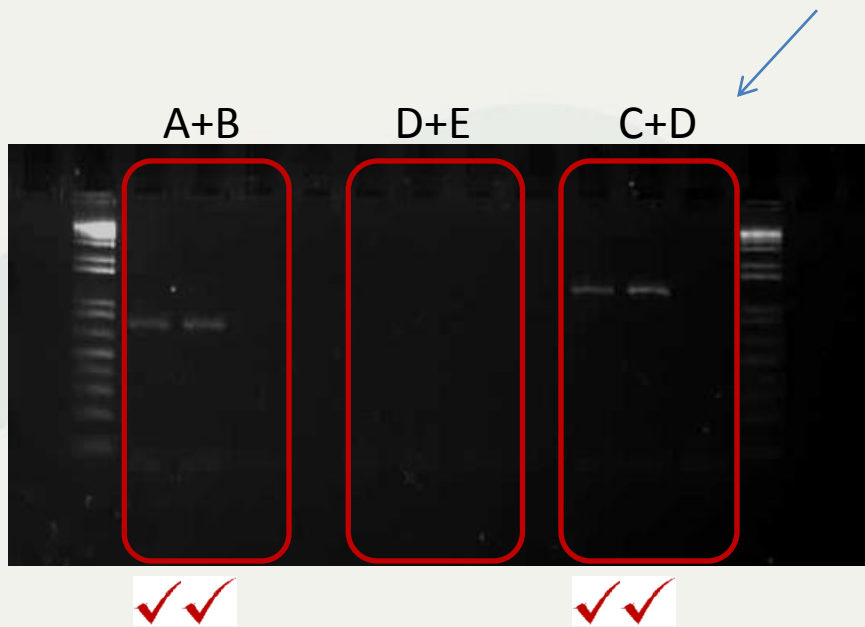
CEU daughter and father in the CEU trio have the pgene in the correct location.

Parent gene id	Name	NA12878	NA12891	NA12892
ENSG00000122565	CBX3	Yes	Yes	Yes
ENSG00000139372	TDG	Yes	No	No
ENSG00000165480	SKA3	Yes	Yes	No



SKA3

Predicted size = ~1200 bp



A + B pgene exists
 E + D no pgene at predicted site (for ≥ 1 chr)
 C + D pgene at predicted site (for ≥ 1 chr)

CEU daughter and father in the CEU trio have the pgene in the correct location.

Parent gene id	Name	NA12878	NA12891	NA12892
ENSG00000122565	CBX3	Yes	Yes	Yes
ENSG00000139372	TDG	Yes	No	No
ENSG00000165480	SKA3	Yes	Yes	No





Future Directions

- Run more product on gel to get prettier pictures (these were 2 uL of 25 uL rxns)
- Get E+D primers to work
 - Temperature gradients with CEU mother
- Sequence PCR products
- What's next?