

1. The complete recessive allele *sd1* conferring the short-stature phenotype is the key to resolving the lodging problem when rice production applied nitrogen fertilization. Please answer the questions below:
- a. Scientists chose a map-based cloning approach to identify the *SD1* gene. The preliminary investigation revealed that the *SD1* gene is on chromosome 1 using an $F_{2:3}$ mapping population from a cross of two rice varieties: Milyang 23 (homozygous *sd1*) and Gihobyeo (homozygous wild-type presenting with the "+" symbol). The plant height (culm length) of Milyang23 was in the range of 60 - 80 cm. The plant height of Gihobyeo and F_1 plants were in the range of 95 - 140 cm. The tables below show all pairs of genotypes of four loci (the *SD1* gene and three RFLP markers) from the same 60 F_2 individuals: A/A genotype is homozygous for Gihobyeo; A/B genotype is heterozygous; B/B genotype is homozygous for Milyang 23. The physical distance between RG109 and RG220 had later validated at approximately 260 kb. What is the valuable information obtained from this preliminary investigation? How did it influence the map-based cloning strategy? What specific plan would you like to do to continue the map-based cloning of this *SD1* gene? (25 points)

<i>SD1</i>	RG109			RG220			RG381		
	A/A	A/B	B/B	A/A	A/B	B/B	A/A	A/B	B/B
+ / +	15	0	0	15	0	0	12	3	0
+ / <i>sd1</i>	0	29	1	0	29	1	0	29	1
<i>sd1</i> / <i>sd1</i>	0	0	15	0	0	15	0	1	14

Critical values of the χ^2 distribution				
df	1	2	3	4
P=0.05	3.841	5.991	7.815	9.488

RG381	RG109			RG220			RG109	RG220		
	A/A	A/B	B/B	A/A	A/B	B/B		A/A	A/B	B/B
A/A	12	0	0	12	0	0	A/A	15	0	0
A/B	3	29	1	3	28	2	A/B	0	28	1
B/B	0	0	15	0	1	14	B/B	0	1	15

- b. Two rice mutants, D51, and D66, derived from γ -ray irradiation on the rice cultivar Calrose, show a short-stature phenotype. Investigation on the genetic relationships between these genes conferring short-stature phenotype used two F_2 populations: one derived from the cross of G33 (with homozygous *sd1* allele) and D51, the other from the G33 x D66 cross. Individual F_2 plants from the G33 x D51 cross showed similar plant height with low phenotypic variation within the whole population. However, individual F_2 plants from the G33 x D66 cross showed distinct plant height and significant phenotypic variation within this F_2 population. Thus, individual F_2 plants in the G33 x D66 cross were classified using two criteria. (1) The plant height of the F_2 individual, including three distinct levels: tall, semi-dwarf, and dwarf. (2) Phenotypic variation within the self-pollinated F_3 progenies of a single F_2 individual: "uniform" means variation of plant height kept in the same level, and "segregating" means variation of plant height appeared in different levels. It resulted in five different phenotypic classes in the G33 x D66 cross, shown in the tables below. Please formulate your hypothesis on the genetic factors conferring short-stature phenotype in these three rice cultivars (G33, D51, and D66). You need to use the Chi-square test to validate your hypothesis. (25 points)

	Plant Number	Plant Height
Parental lines		
G33	63	88 ± 18.1 cm
D51	60	90 ± 14.1 cm
D66	59	96 ± 11.4 cm
Calrose (wild type)	47	108 ± 16.1 cm
Crosses		
F2 of G33 x D51	66	86 ± 7.6 cm
F2 of G33 x D66	102	99 ± 171.8 cm

Class in the G33 x D66 cross	Observed F_2 Plant Number
Tall, uniform	10
Tall, segregating	49
Semi-dwarf, uniform	10
Semi-dwarf, segregating	29
Dwarf, uniform	4

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2. Please fill the blank space with one single alphabet ("a" to "k" only) and write the 8 alphabets on answer sheet in the order of appearance (1 point each, total 8 points)

The full chemical name of DNA is _____. A single strand of a DNA molecule consists of a chain of subunit called _____, each subunit is made of the sugar _____ connected to an inorganic _____ and to one of four _____, the bonds joining one subunit to another are covalent _____. DNA synthesis requires key enzymes called _____ which add subunits successively onto the _____ end of a growing strand.

- a. nucleotides
- b. hydrogen bonds
- c. deoxyribose
- d. nitrogenous bases
- e. 3'
- f. phosphate group
- g. deoxyribonucleic acid
- h. DNA recombinase
- i. phosphodiester bonds
- j. DNA polymerase
- k. 5'

3. Please select one answer from "a" to "h" to correspond to condition 3.1 to 3.4 respectively

3.1 To examine if the mutations arise from random events *or* in response to environmental signal (2 points)

3.2 To identify if a compound is a potential carcinogen (2 points)

3.3 To break the sugar-phosphate backbone and thereby split DNA into smaller piece (2 points)

3.4 To diagnose if someone could potentially inherit mutant allele causing fragile X syndrome (the excess amplification of a CGG base) (2 points)

- a. A complementation test
- b. Expose to ultraviolet (UV) radiation
- c. Examine the number of thymine dimers
- d. The Luria-Delbruck fluctuation experiment
- e. Expose to X-ray
- f. Analyze the number of particular trinucleotide repeats
- g. The Ames test
- h. The Hershey-Chase Waring blender experiment

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4. Your professor studying genes influencing stamen number in the plant *Arabidopsis thaliana* identified six independent recessive mutations that resulted in plants that had abnormal stamen number rather than six in wild type. One of your lab mates started to perform complementation tests with these mutants, but some of the tests could not be completed because of lacking sufficient time. The results of the tests that were finished are shown in the table the follows.

	1	2	3	4	5	6
1	-	+	-	?	+	?
2		-	?	?	?	-
3			-	-	?	?
4				-	?	?
5					-	+
6						-

- 4.1 What experiment was performed to conclude these six mutants are recessive mutations? (2 points)
- 4.2 Please describe how the experiment was done so the results could be used to fill in individual boxes with a plus (+) or a minus (-) (2 points)? What experimental results does + and - represent? (4 points)
- 4.3 Assuming no complications, what do you expect for the results of the unperformed experiments labelled with “?”, please draw the same table in the answer sheet and complete the table by placing a + or a - to replace “?” in each box (10 points)
- 4.4 How many genes are represented among this collection of mutants? (4 points) Which mutations are in which gene groups? (4 points)
5. Mutations that alter the nucleotide pairs of DNA can modify any of the steps or products of gene expression.
- (a) Below are four types of mutation which occurs in a gene’s coding sequence, please describe the effect of each mutation on gene’s product
1. Silent mutation (1 points)
 2. Missense mutation (1 points)
 3. Nonsense mutation (1 points)
 4. Frameshift mutation (1 points)
- (b) What is “gain-of-function” allele? Why are they almost always dominant to wild-type alleles? (4 points)

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