Possible Complications in Mendel's Pea Experiment

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Some Background...

- In 1856, Gregor Mendel began his flagship experiments on Common Garden Edible Pea (*Pisum Sativum*) to observe the inheritance patterns of the plant's 7 distinct traits:
  - Plant Height
  - Flower Colour
  - Pod shape
  - Pod Colour (unripe)
  - Seed Shape
  - Seed Coat Tint
  - Flower Location.
- After extensive studies, Mendel proposed his 3 namesake laws of inheritance: **Law of Segregation, Law of Independent Assortment** and the **Law of Dominance.**

Mendel's Laws of Inheritance

- The **Law of Segregation** states that each diploid individual has a pair of alleles (copy) for a particular trait and each parent passes an allele at random to their offspring via meiosis.

- The **Law of Independent Assortment** states that the alleles of two (or more) different genes get sorted into gametes independently of one another. In other words, the allele, a gamete receives for one gene does not influence the allele received for another gene.

- The **Law of Dominance** states that when two alleles of an inherited pair are in heterozygous form, then the allele that is expressed is dominant whereas the allele that is not expressed is recessive.

The Controversy!

- In 1936, R. A. Fisher theoretically and statistically reanalysed Mendel's experiments.
- He concluded that Mendel's results with basic phenotypic ratios (3:1) were far too accurate when compared to his own estimation of error-corrected results (3.009:1).
- This meant that:
  - either Mendel did not account for any errors (absolute or relative),
  - or the published data was manipulated / falsified,
  - or Mendel simply got lucky with his observations.

Mendel's Luckiness Parameters

❖**Choice of Plant:** His choice of the Garden Pea was indeed a paramount and crucial decision. Some of the salient features of the Pea plant were:

- The plant reproduces sexually.
- The flowers of this plant are bisexual.
- The flowers are also self-pollinating, and thus self and cross pollination can easily be performed.
- The plants have a high fecundity.
- The different physical characteristics of the plant were easy to recognize and study.
- The plants have a short life span and hence they are easier to maintain.
- The plant has a rapid growth rate.
- Majority of the hybrids are viable.
- It is relatively easy to generate pure lines.

Mendel's Luckiness Parameters

- **Sampling Bias:** If the sample plants that Mendel chose for his experiments were biased towards a specific condition (eg. adaption to the location's temperature, mating of neighbouring plants instead of random mating (due to feasibility), etc), then it may have been possible that his observations were biased and were not universally valid for all pea plants.

- **Sample Size:** It is said that Mendel used ~ 28000 pea plants for his experiment. However, the principles of population genetics were not known back then. Hence, it seems unlikely that Mendel had a defined logic behind his choice his sample size.

Mendel's Luckiness Parameters

- **Occurrence of Phenocopies:** Phenocopy is the phenotype showing features characteristic of a genotype other than its own, but produced environmentally rather than genetically. Mendel would have most probably been able to recognise phenocopies within his dataset since they are non-heritable. However, it would have definitely complicated his analysis to some extent. Furthermore, the obtained phenocopies would be inviable for further analysis due to their incapabilities of displaying their true phenotypes.

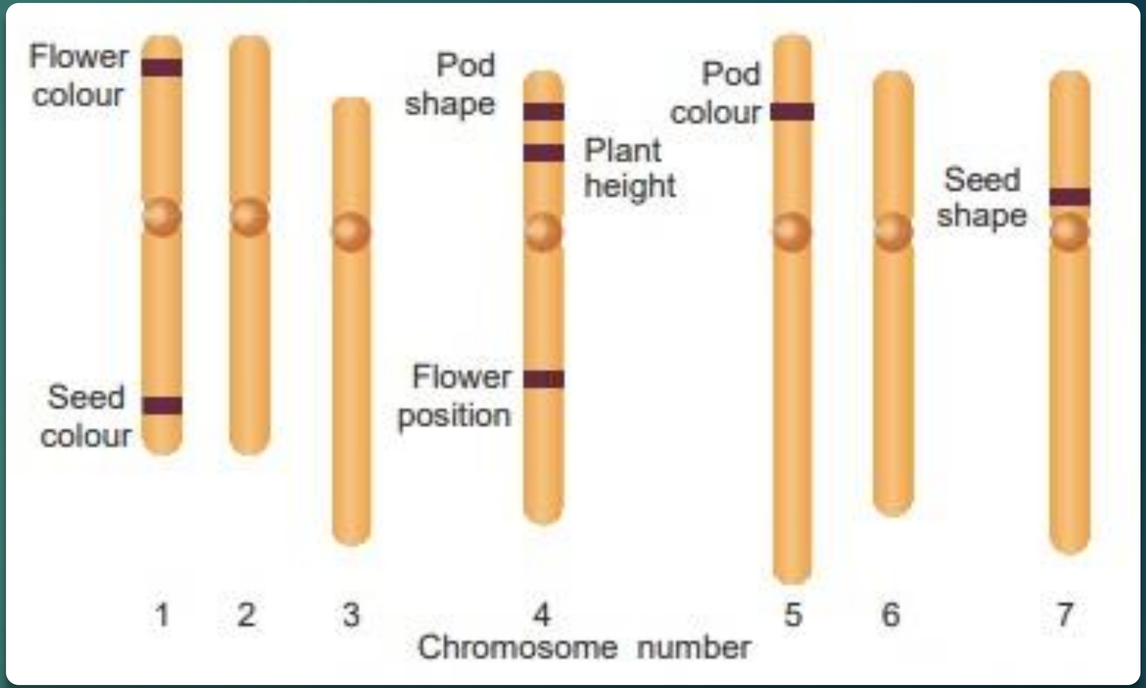
Mendel's Luckiness Parameters

- **Preservation of Dataset:** Among all the pea plants that Mendel would have bred and observed, there is a definite possibility that some of the plants would be eaten by Herbivores, some others could have been damaged by pests, parasites, viruses, natural calamities and anthropogenic interference, and yet others could have displayed phenocopies such as malnutrition or stunted growth due to non-uniform fertilization and care. All these factors contribute to a loss in the data that could have otherwise provided much more accurate observations.

- **Ploidy of the Plant:** The greater the ploidy of the organism, the greater its complexity (more combinations of genotypes created), and thus making it difficult to analyze a larger number of phenotypes available in the dataset. With that being said, a haploid organism does not provide much scope for genetic variation, hence a diploid plant like the pea is the most suitable candidate for analysis.

Mendel's Luckiness Parameters

- **Degree of Linkage:** After the dicovery of chromosomes as the package of genetic material, it was assumed that the genes controlling the 7 traits (observed by Mendel) were on 7 distinct pea chromosomes. However, at a later date it was found that they were spread across 4 distinct chromosomes with the genes controlling plant height and pod shape being particularly close to each-other. Even though the rest of the genes may have been far apart to **assume** minimal linkage between them, the afore-mentioned genes were close enough to exhibit significant linkage. This meant the violation of the Law of Independent Assortment.

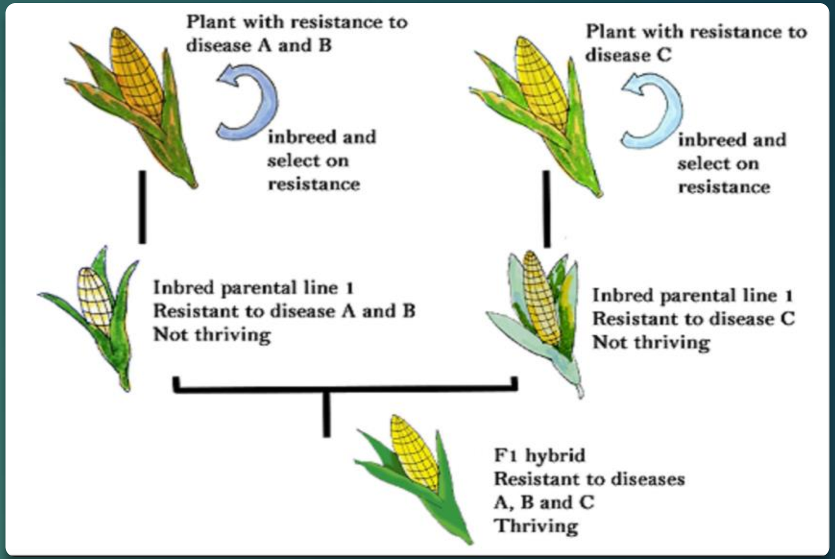


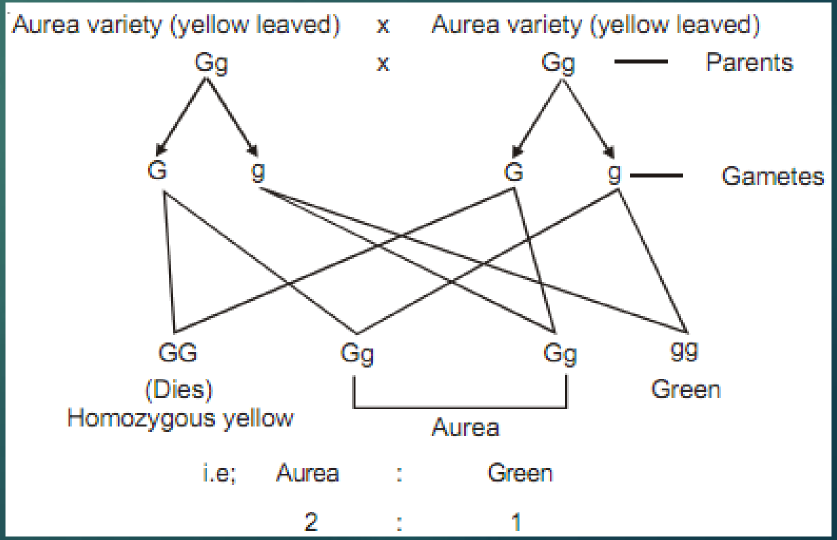
Mendel's Luckiness Parameters

- **Intra-genic / Inter-allelic Actions:** These are interactions between two alleles within a single gene. There are several types of actions which could have complicated Mendel's studies. Some of them are:
  - **Incomplete Dominance:** The dominant gene does not completely mask the expression of the recessive gene resulting in an intermediate phenotype shown by the heterozygote. If Mendel had chosen a trait which shows incomplete dominance (such as the flower colour of Rose), he would have never coined the Law of Dominance as it is directly violated.
  - **Codominance:** 2 or more alleles in a gene are equally expressed. This results in the expression of all the dominant alleles in the phenotype of the heterozygote. This too violates the Law of Dominance as can be seen in the flower colour of the Rhododendrons.

Mendel's Luckiness Parameters

- **<u>Over-dominance</u> / <u>Heterozygous Advantage</u> / <u>Hetorosis</u> / <u>Hybrid Vigour</u>:** Heterozygote produces a phenotype more extreme or better adapted than that of the homozygote. As can be seen in the increase in fecundity of Corn plants, Overdominance (if present) would have definitely skewed the ratios of the subsequent generations towards a more heterozygotic population.
- **<u>Lethal Allelism</u>:** Alleles that cause the death of the organism that carries them. Both dominant as well as recessive lethals could have posed a significant threat to Mendel's experiment as it would have created a monoallelic population over time. The insufficiency of chlorophyll in some Snapdragons (due to lethals) could have been the perfect recipe for disaster in Mendel's experiment.





Mendel's Luckiness Parameters

- **Multiple Allelism:** Three or more alternative forms of a gene (alleles) that can occupy the same locus. While Mendel was adept at dealing with 2 allele systems, even if one of the traits that he chose would have shown multiple allelism (like the 4-allele controlled trait of flowering onset in the Pea plant), the genotype and phenotype ratios could have skewed enough for him to not coin the laws of inheritance.

- **Inter-genic / Non-allelic Actions:** Development of single character is due to two or more genes affecting the expression of each other in various ways. The majors complication in these actions is the skewness of phenotypic ratios and the large number of phenotypes for analysis. These can be:
  - **Pseudo-Overdominance:** Heterozygous forms of 2 genes (that are very close to each other on the same chromosome) is more extreme or better adapted than that of the homozygotes of the 2 genes. The phenotype analysis complications would be similar to that of the heterozygous advantage.

Mendel's Luckiness Parameters

- **Duplicate Gene Action:** Two loci's gene products have the same (redundant) functions within the same biological pathway. Eg. awn character in Rice.
- **Complementary Gene Action:** Loss of function of either *A* or *B* gene function has the same phenotype as the loss of function of both genes. It means that the functions of both genes work together to produce a final product. Eg. flower colour in Sweet Pea plant (*Lathyrus odoratus*).
- **Supplementary Gene Action:** Dominant allele of one gene is essential for the development of the concerned phenotype, while the other gene modifies the expression of the first gene. Eg. grain colour in Maize.
- **Inhibitory Gene Action:** One dominant inhibitory gene prevents the expression of another dominant gene. Eg. Anthocynin pigmentation in Rice.
- **Polymeric Gene Action:** When two genes govern any character separately, their effect is equal but when both the genes are present together, there phenotypic effect is increased or raised as if the effects of the two genes were additive or cumulative. It is notable in this case that both the genes show complete dominance. Eg. fruit shape of Summer Squash.
- **Dominant Epistasis / Masking Gene Action:** Dominant allele at one locus may mask the phenotype of a second locus. Eg. fruit colour in Summer Squash.
- **Recessive Epistasis:** When the recessive allele of one gene masks the effects of either allele of the second gene.

Mendel's Luckiness Parameters

- **Non-discrete / Quantitative Traits:** Traits that are governed by more than 2 genes (polygenic traits) give a continuum of phenotypes such as leaf colours, leaf length, etc. They usually form a continuous variation curve with a Gaussian Distribution. Had Mendel chosen one of these traits, it would have been impossible for him to formulate any conjectures because of the sheer number of phenotypes to observe and analyze.

- **Environmental Penetrance and Expressivity:** Genetic (pleiotropic) and environmental impacts can cause an incomplete penetrance and variable expressivity which in turn could have caused difficulty in observations, analysis and a loss of dataset.

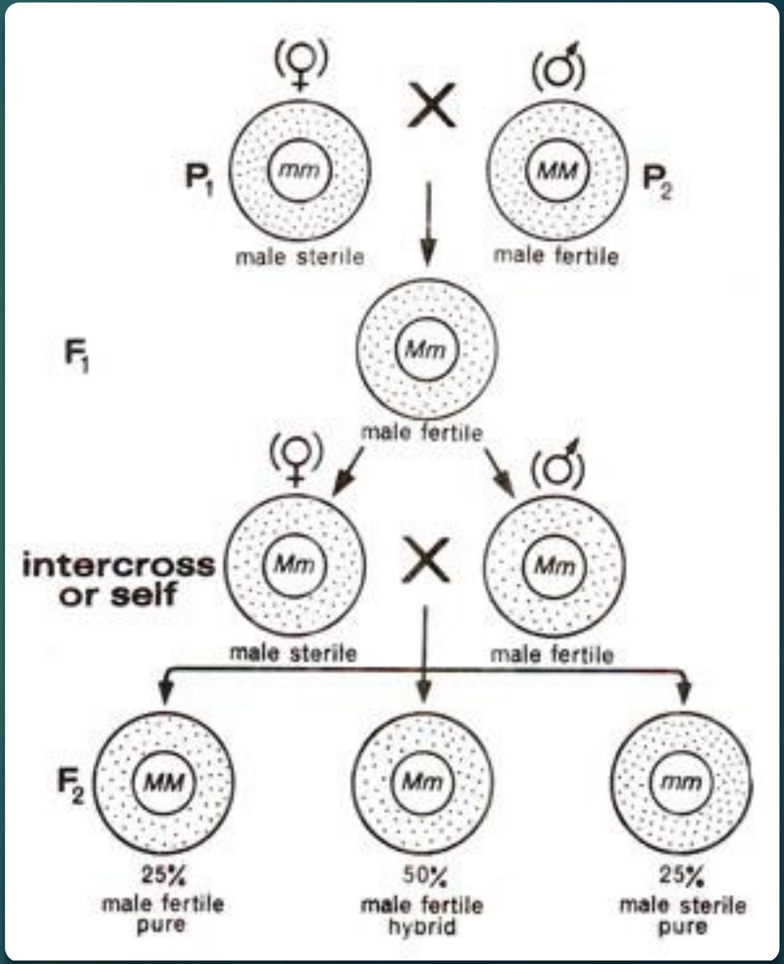
Mendel's Luckiness Parameters

- **Effects of Pleiotropy:** One gene influences two or more seemingly unrelated phenotypic traits. As a matter of fact, 3 such traits were observed by Mendel in his experiments (presence of seed-coat colouration, presence of flower colour and presence of coloured leaf axils), however he could not theorize these abnormalities. This definitely confused Mendel and could have possibly delayed his deductions of the inheritance patterns.

- **Consideration of Sex-linked traits:** According to recent studies, sex-linked house-keeping genes were revealed in the White Campion (*Silene Latifolia*). This definitely points towards existence of observable sex-linked traits in plants. Had Mendel to have chosen a unisexual plant and have encountered such a trait, it would have definitely complicated his formulation of laws.
  However, knowing Mendel, he would have perhaps figured out its sex-linked properties.

Mendel's Luckiness Parameters

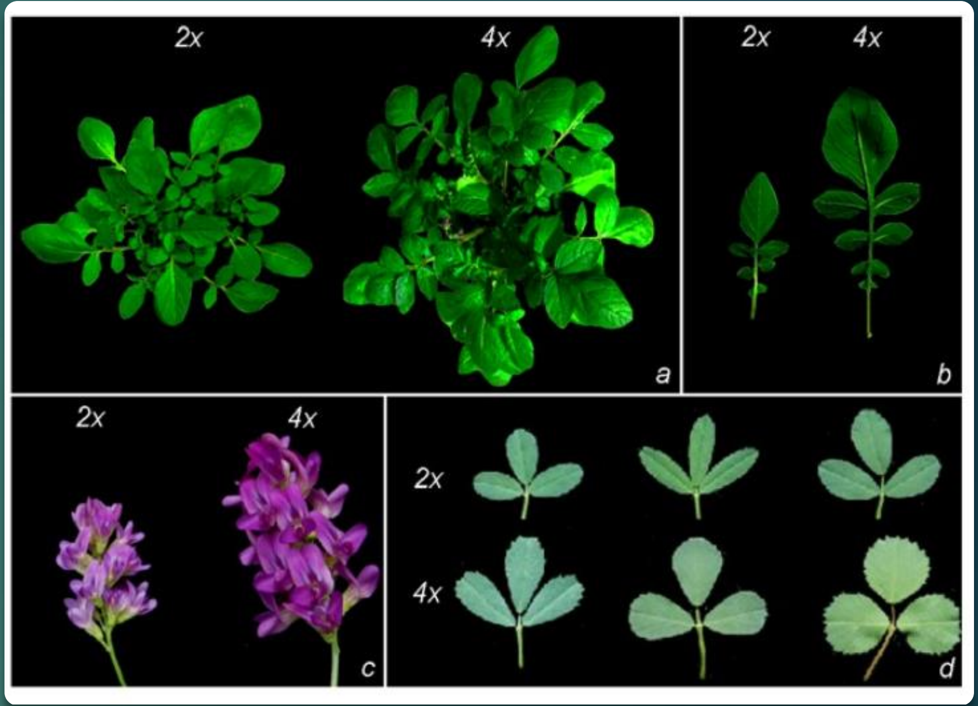
- **Cytoplasmic Inheritance:** This mode of inheritance is possible through the mitochondria, chloroplast, endobacteria and viruses. It can be either uniparental or biparental depending on its genetic origins. Since it is an extranuclear inheritance, it is completely non-Mendelian in nature and hence nearly impossible to figure out without microscopic and lab-experimental aids. In this case, Mendel was definitely lucky to have not chosen such a trait, which are readily existent as examples of cytoplasmic male sterility in several plant crops such as Maize.



- **Heritable Mutations:** This factor should not have troubled Mendel to a large extent, since mutations get fixed into the phenotype, thereby permanently altering it.

Mendel's Luckiness Parameters

- **Effects of Polysomy / Polyploidy :** Several known examples of polysomy / polyploidy in plants (such as Mustard, Eyebright, Dallas Grass and the entire Liliceae family) suggest that Mendel could have stumbled upon any of them as a model organism for his experiments. Phenotypic expression of polyploids could definitely be eliminated as semi-hereditary, however it would have taken some extra effort on Mendel's part to do so.



- **Action of Transposons:** As the action of a transposon is unpredictable, it can lead to a very high phenotypic plasticity. Since Mendel was a pioneer in his field, encountering something as complex as a transposon would have indeed lead his research to a dead-end.

Conclusion

- Looking at all the above complications that could have arisen in Mendel's studies, it can be said that Mendel was surely a lucky researcher.
- However, as mentioned earlier, some of those complications did cross paths with Mendel, but he tactfully managed to improve the situation by manipulation of his results (probably) and change his model organisms from time to time untill he settled on the Garden pea plants. His choice of the Pea plant could be said as either the luckiest thing that happened to him OR it was his smartest move (depending on the perspective).
- With this being said, there is no denying the fact that without Mendel's revolutionary accomplishments, the scenery of Evolutionary Genetics would have been starkly different today. So despite Mendel's possible ethical and academic shortcomings, the world is in his debt to coin the field of "Mendelian Genetics"!