*In this section of* **Resonance**, *we invite readers to pose questions likely to be raised in a classroom situation. We may suggest strategies for dealing with them, or invite responses, or both. "Classroom" is equally a forum for raising broader issues and sharing personal experiences and viewpoints on matters related to teaching and learning science.*

## Teaching and Learning Genetics with *Drosophila*
## 2. Mutant phenotypes of *Drosophila melanogaster*

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**The first part of this series [1] looked at why the fruitfly *Drosophila* is such an excellent eukaryotic model system for genetic studies. Of the many important attributes of this fly, which have earned it the title *'Cinderella of Genetics'*, the availability of innumerable number of mutant genetic stocks of *D. melanogaster* is an extremely useful and important one. These different mutants are of immense help to study various aspects of inheritance, as we shall see in this and subsequent articles.**

### What is a Mutant Phenotype?

Each and every observable character or trait of an individual (i.e. its phenotype) is controlled by specific gene(s). Whenever a gene undergoes a change, the message of the gene is altered, and this will have an effect on the product and, therefore, the function of that gene. As a consequence of this, the phenotype, which was under the control of this gene is going to be changed. This altered phenotype is called a 'mutant phenotype', and the event of the gene undergoing such a change is termed 'mutation'. A mutation, thus, is a heritable change in the genetic material. Every gene can undergo mutation, but different mutations have

different types of effects on the phenotype and, moreover, they have different mutation rates. In any species, including humans, every individual is a carrier of mutations for one or the other gene. For instance, a few mutant phenotypes in man are curly hair, baldness, eye colours, albinism, sickle-cell anaemia, xeroderma pigmentosum, polydactyly, hemophilia, night blindness etc. Similarly, mutations and mutant phenotypes are seen in all species including *Drosophila*, maize, worms, frogs, mouse etc. Mutations occur spontaneously in nature, but at a very low rate. Chemical, physical and biological agents can also induce mutations in the laboratory. The father of genetics, Gregor Mendel, exploited the normal and mutant forms of the pea plant and gave us important generalizations concerned with the norms of inheritance.

## What is so Special about Mutant Phenotypes of *D. melanogaster*?

Ever since T H Morgan introduced *Drosophila melanogaster* as a model system for genetic research in 1909, countless numbers of geneticists around the world have exploited this system. During these nine decades, hundreds of mutations have been isolated and definite specific stocks (true-breeding populations) of these

**Box 1. Dominant and Recessive Genes**

Based on how its expression is affected by the other copy of the gene in a diploid individual, a mutation is termed either dominant or recessive. A mutation is said to be 'dominant' if it is expressed in the heterozygous condition. That is, one of the homologous chromosomes carries the mutant form of the gene (allele), while the other has the normal (wild type) allele. A 'recessive' mutation is one that needs to be in homozygous state for its expression. That is, the same mutant allele has to be present on both the homologous chromosomes for the mutant phenotype to be expressed. On the other hand, one dose of recessive mutation on the X-chromosome of male *Drosophila* is expressed, because in males the chromosome Y is not completely homologous to its counterpart – X-chromosome, and does not carry the corresponding alleles of the X-chromosome. This situation in which males are functionally homozygous for X-chromosome genes, is called 'hemizygosity'. However, in females, the same recessive mutation on the X-chromosome needs to be in 'homozygous' state for expression. Of course, some mutations lie between these extremities, and are termed semidominant or codominant. In such cases, an individual heterozygous for the mutant allele and the wild type allele will have a phenotype intermediate between the normal and mutant phenotypes.

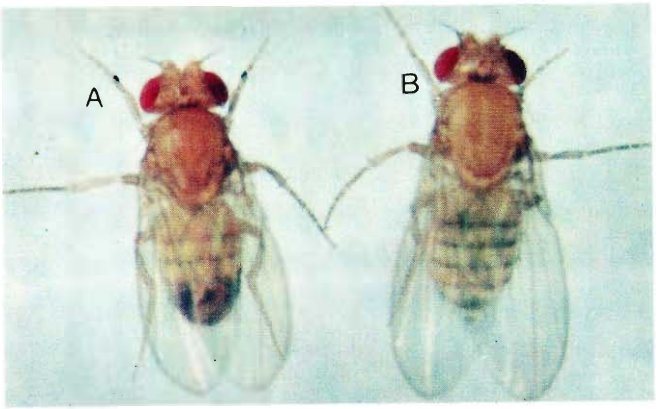



*Figure 1. Normal Male (A) and Female (B) of D. melanogaster. Features of these flies, particularly of eye, body colour and wings have to be compared with the mutant phenotypes presented in Figures 2 to 8.*

mutant phenotypes have been established. As of now, over 4000 genes with their various alternative mutant forms have been recognised in *D. melanogaster* (Lindsley and Zimm, 1992). The Drosophila Stock Centre at Mysore has about 2000 genetic stocks of *D. melanogaster*; each stock breeds true for a particular mutant phenotype(s). For instance 'white eye' stock means a homozygous strain which breeds true for this phenotype. Such genetically defined mutant strains are a prerequisite for a systematic analysis of inheritance of characters and for the genetic dissection of phenotypes. The genetic repertoire of *D. melanogaster* includes a wide variety of mutants of importance to the understanding of genetics, development, behaviour and cell biology. For most of the mutants, sufficient background information ranging from its final morphological manifestation to its actual functions at cellular and molecular levels is available. Thus, a wealth of mutant stocks and a solid body of knowledge and literature are at our disposal. Today it is possible to engineer specific genetic strains of *D. melanogaster* with desired gene combinations, almost at will, to suit the nature of investigation and/or to test a hypothesis. Therefore a *Drosophila* worker today starts at a relatively advanced stage in terms of the type of question he can ask of his material. In view of these benefits, even though mutants are available in other species of *Drosophila* and non-drosophilid systems, including humans, none of them can match *D. melanogaster* in its utility and suitability as a test system for genetic studies.

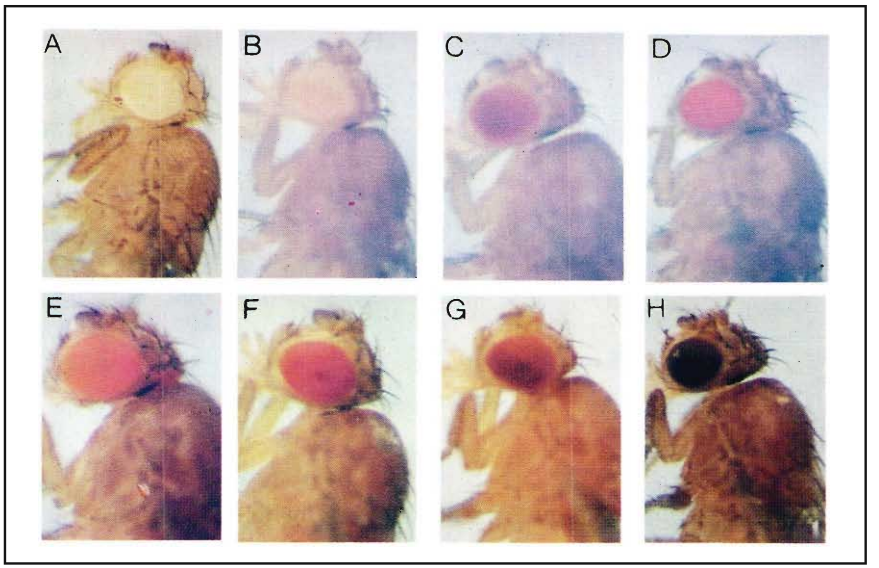



*Figure 2A-H.*
*Eye colour mutants:*
*(A) white*
*(B) white apricot*
*(C) brown*
*(D) scarlet*
*(E) cinnabar*
*(F) vermilion*
*(G) rosy*
*(H) sepia.*

**Description of a Few Mutants of *D. melanogaster***

Many mutant stocks of *D. melanogaster* for different characters in various combinations are available at Drosophila Stock Centres. We have here selected a few representatives for description.

(A) ***Eye Mutations (Figure 2A-H)***: The normal eye colour of *D. melanogaster*, is red (*Figure* 1A-B) due to colored pigments. In a mutant these pigments are absent and, hence, the eye appears white. In other mutants, depending on the presence or absence of one or the other pigment, the eye shows different shades of colour. The mutant phenotype and the respective gene symbol in brackets for a few eye mutants are as follows: *white (w)* (*Figure* 2A), *white apricot* ($w^a$) (*Figure* 2B), *brown (bw)* (*Figure* 2C), *scarlet (st)* (*Figure* 2D), *cinnabar (cn)* (*Figure* 2E), *vermilion (v)* (*Figure* 2F), *rosy (ry)* (*Figure* 2G), *sepia (se)* (*Figure* 2H).

***(B) Wing Mutations (Figure 3A-I):*** *D. melanogaster*, a dipteran, has a pair of wings, which have a definite shape and structure, as well as orientation on the body (*Figures* 1A-B; 3D). Because of mutations in the genes, which determine these features, many wing mutants are available. Wings may be cut to points and edges scalloped – *cut (ct)* (*Figure* 3A); wings can be extremely

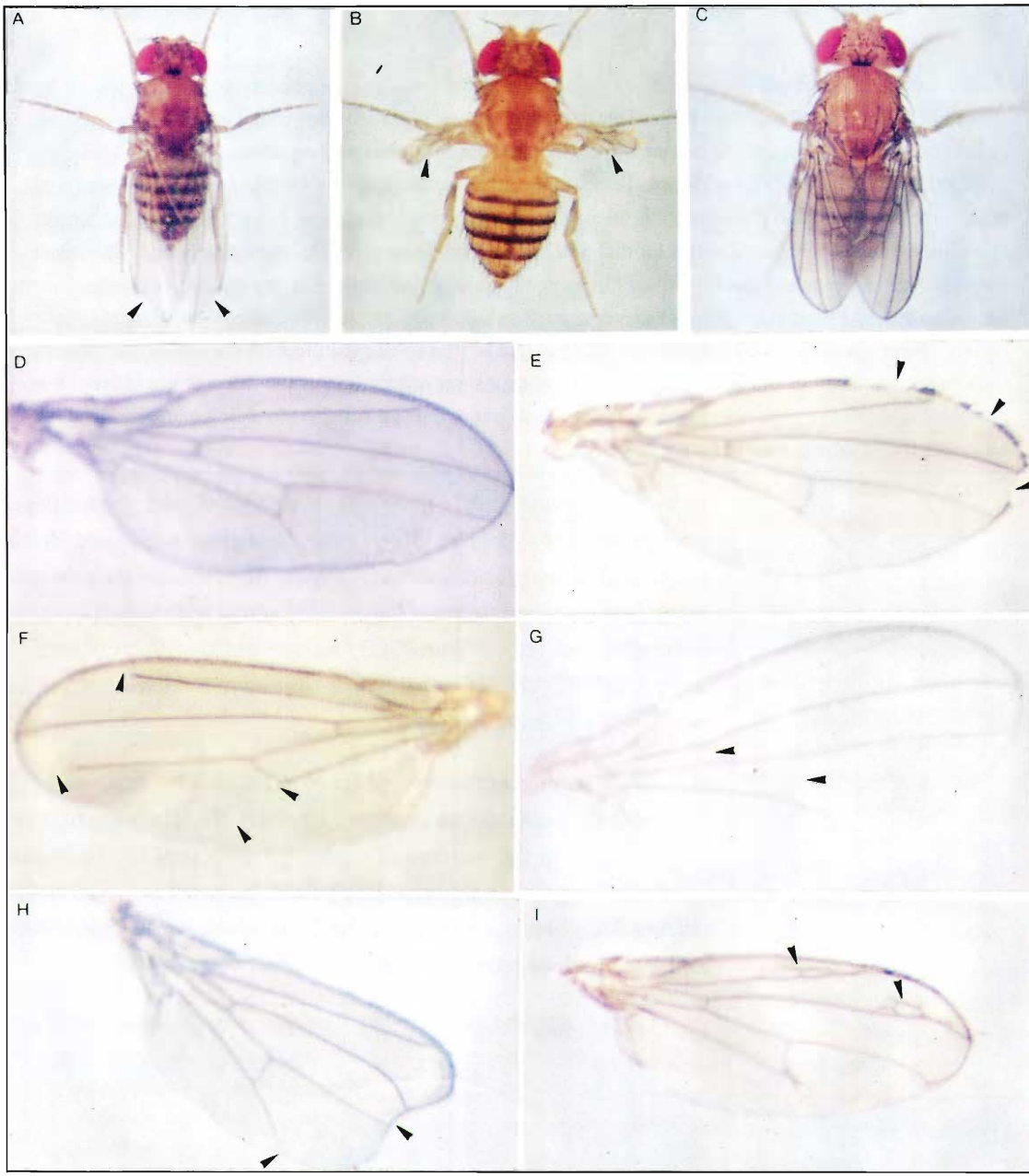



***Figure 3A-I. Wing mutants: (arrowhead points the mutant region of the wing). (A) cut wing (B) vestigial (C) microptera (D) normal wing (E) scalloped (F) veinlet (G) crossveinless (H) dumpy (I) plexus.***

**Box 2. Genes and Symbols.**

Traditionally, the name and the symbol of a gene are derived from its mutant phenotype, and they have to be written in italics. Usually, the first letter of the mutant phenotype is used for the symbol of the concerned gene. A lower case letter is used if the mutant is recessive, and for the corresponding normal dominant allele, the same symbol will have a '+' superscript. For example, in *D. melanogaster* $w$ = white eye, $w^+$ = red eye (wild type). On the other hand, if the mutant is dominant, the upper case letter is used to name the gene. Example: $Cy$ = curly wing, $Cy^+$ = normal wing. Further, if the same letter is involved for two different mutations, then for one of them two letters have to be used to name the gene. Example: $w$ = white eye, $wg$ = wingless, $v$ = vermilion eye, $vg$ = vestigial wing. Whenever two letters are employed, care should be taken to avoid a space between them. Sometimes the genes are pleiotropic, that is, the same gene influences more than one trait. In such cases, the name of the gene is derived from the most prominent trait of the affected characters. For instance the white eye gene not only determines the colour of the eye but also the malpighian tubes and the testes but the letter $w$ is used as its symbol.

reduced – *vestigial (vg)* (*Figure* 3B); wings small and spoon like – *microptera (mp)* (*Figure* 3C); wing margins scalloped with thickened veins – *scalloped (sd)* (*Figure* 3E); wing veins do not reach margins – *veinlet (ve)* (*Figure* 3F); wings without crossveins – *crossveinless (cv)* (*Figure* 3G); wings obliquely truncated – *dumpy (dp)* (*Figure* 3H); and wings that have a network of extra veins – *plexus (px)* (*Figure* 3I).

***(C) Body colour mutations (Figure 4A-C):*** The normal body colour of *D. melanogaster* is grey (*Figure* 1A-B). Here again, due to mutations in the concerned genes, the colour of the body can change giving rise to mutants like yellow body colour – *yellow (y)* (*Figure* 4A); black pigment on the body – *black (b)* (*Figure* 4B); and body colour shining black – *ebony (e)* (*Figure* 4C).

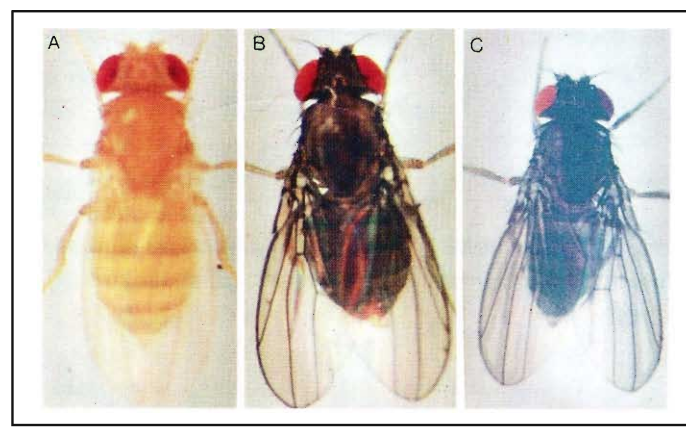



***Figure 4A-C. Body colour mutants: A) yellow (B) black (C) ebony.***

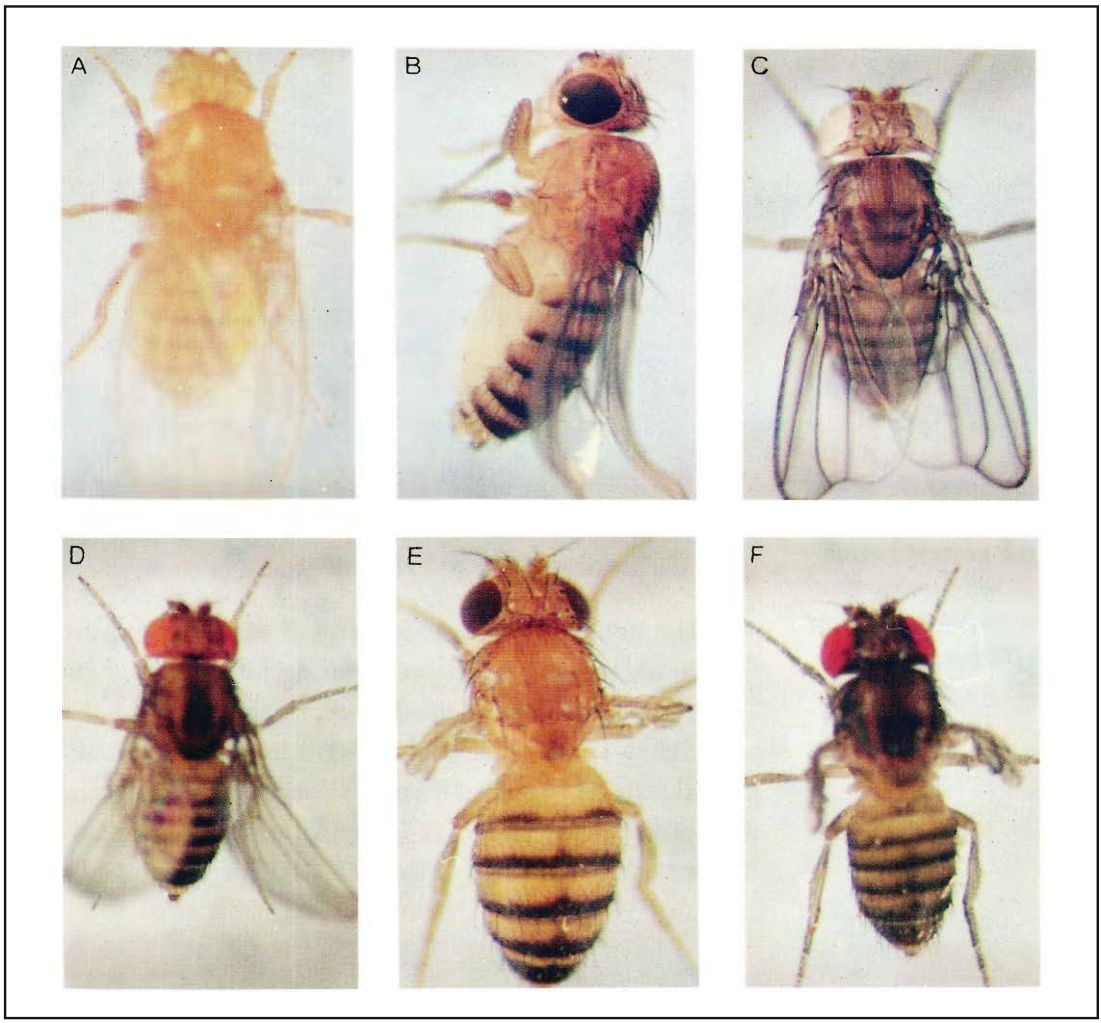



***Figure 5A-F. Multiple mutants: (A) yellow body, white eye (B) sepia eye, curled wing (C) white eye, dumpy wing, black body (D) curled wing, ebony body, dark stripe on thorax, ruby eye (E) sepia eye, vestigial wing (F) ebony body, vestigial wing.***

***(D) Multiple Mutations (Figure 5A-F):*** Genetic strains of *D. melanogaster* with more than one mutant phenotype are also available. For example, yellow body with white eyes (*y w*) (*Figure* 5A); sepia eye colour with curled wing (*se cu*) (*Figure* 5B); dumpy wing, white eyes and black body (*dp w b*) (*Figure* 5C); curled wing, ebony body, stripes on the thorax, eye colour ruby (*cu e sr ca*)(*Figure* 5D);sepia eye colour with vestigial wing (*se; vg*) (*Figure* 5E); ebony body with vestigial wing (*e; vg*) (*Figure* 5F).

In these multiple mutant stocks, if the concerned genes are within the same chromosome or a linkage group, a space has to be given between symbols of two different genes. On the other

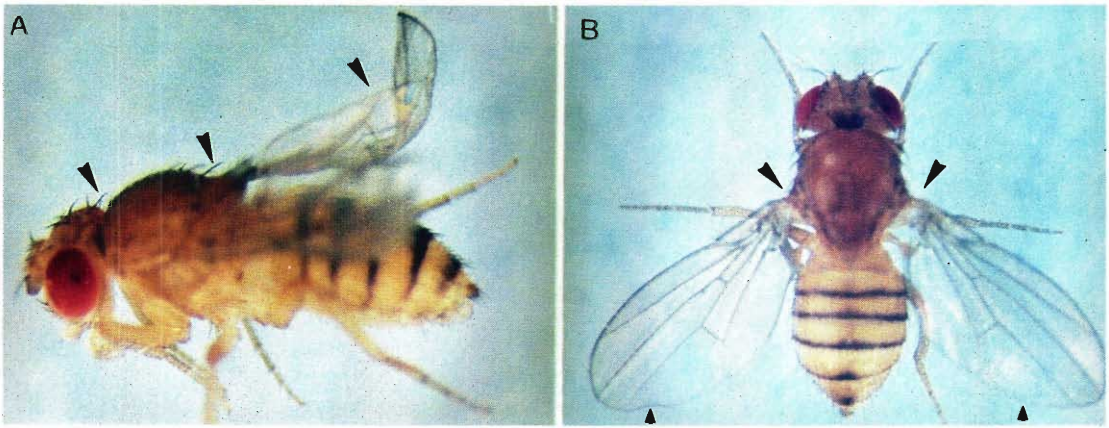



*Figure 6A-B(top). Dominant mutants: (arrowhead indicates the mutant region). (A) Curly wing, short stumpy, Bristles (Bl) Wings held at 45°–Dichaete; narrow wings – Lyra.*

hand, if the mutant genes of the stock are on different linkage groups, then between such gene symbols, punctuation in the form of a semicolon has to be introduced.

***(E) Dominant Mutations (Figure 6A-B and 7A-D):*** Mutations thus far discussed are recessive to normal phenotypes; hence the lower case letters are adopted to represent the symbol of the gene. However, there are mutations which are dominant over normal features. Some of them are *Curly* wing (*Cy*) (*Figure* 6A), short stumpy *Bristles (Bl)* (*Figure* 6A), wings are held at 45° to the body – *Dichaete (D)* (*Figure* 6B), lateral margins of wings

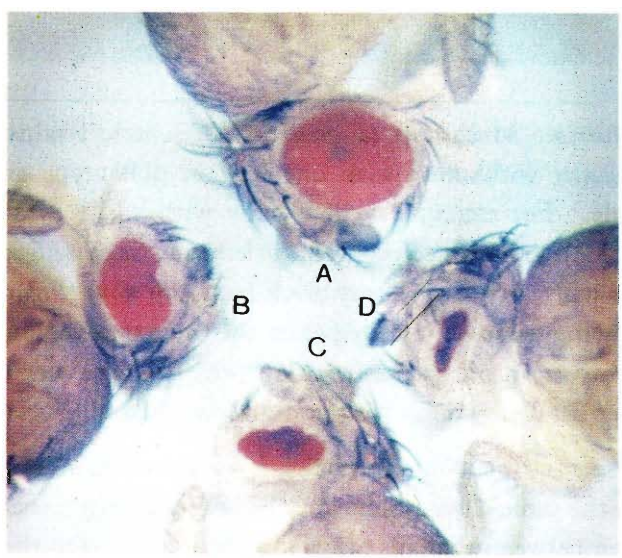



*Figure 7A-D. Normal and bar eye phenotypes compared. (A) Normal eye (B) Bar – kidney shaped (C) Bar – reduced (D) Bar – highly reduced eye.*

reduced giving narrowed shape – *Lyra (Ly)* (*Figure* 6B). An additional feature of these is that each one of them is lethal in homozygous state, meaning that flies of genotypes like *Cy/Cy*, *Bl/Bl*, *D/D*, and *Ly/Ly*, do not survive. Hence, these mutants have to be maintained in heterozygous condition, such as *Cy/Cy*$^+$, *Bl/Bl*$^+$, *D/D*$^+$, *Ly/Ly*$^+$. This does not, however, mean that all dominant mutants are lethal in homozygous condition. One of the celebrated dominant mutants is the *Bar (B)* eye in *Drosophila*, and it reduces the size of the eye. *Figure* 7A-D presents a comparative picture of normal size eye (*Figure* 7A), with those of bar eyes, namely kidney shaped eye (*Figure* 7B), reduced eye (*Figure* 7C) and highly reduced eye (*Figure* 7D).

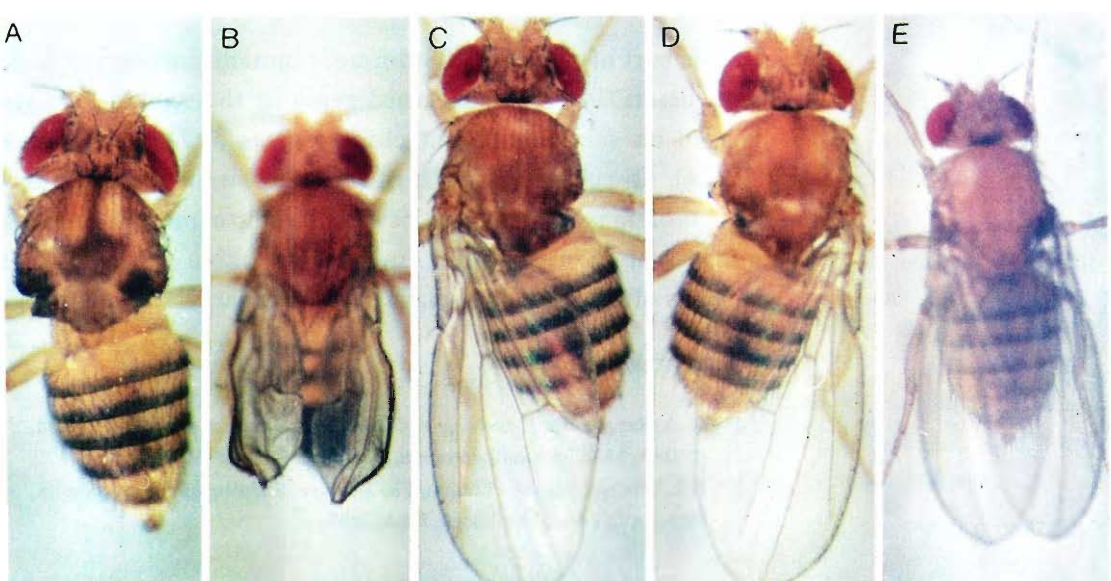
Most of the mutants enjoy total penetrance and expressivity (see *Box* 3). For instance in the white eye stock, the eyes of all the individuals are uniformly white. On the other hand, there are a few mutants whose penetrance and expressivity vary in different individuals of a stock. For example in the wingless stock, all the individuals are homozygous for the mutant allele, *wg(wg/wg)*. In

**Box 3. Penetrance and Expressivity**

Differences in environmental conditions or in genetic backgrounds may cause individuals, which are genetically identical at a particular locus, to exhibit different phenotypes.

The ability of a given gene or gene combination to be expressed phenotypically is called *penetrance*. A trait, though penetrant, may be quite variable in its expression. The degree to which a penetrant genotype is actually expressed is called *expressivity*.

***Figure 8A-E. Phenotypes of the wingless stock. (A) without wings (B) under developed wings (C) right wing only (D) left wing only (E) with both wings.***



**Box 4. Genes and Chromosomes**

*D. melanogaster* has four pairs of chromosomes. The following list indicates the chromosome on which the genes of the mutants described in this article are located. (*These mutants are not described here).

| Chromosomes | Genes | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| X | *y* | *w* | $w^a$ | *cv* | *ct* | *v* | *sd* | *B* | | | |
| 2 | *Cy* | *dp* | *wg* | *b* | *Bl* | *cn* | *vg* | *px* | *bw* | | |
| 3 | *mp* | *ve* | *se* | *D* | *Ly* | *st* | *cu* | *ry* | *sr* | *e* | *ca* |
| 4* | *ci* | *ey* | | | | | | | | | |

this stock one can see flies without both wings (*Figure* 8A) where penetrance is followed by complete expressivity; also flies with incomplete wings (*Figure* 8B); with left wing only (*Figure* 8C); with right wing only (*Figure* 8D) where the penetrance of the mutant gene is followed by different degrees of expressivity; and also flies with both wings (*Figure* 8E) where the penetrance is zero and therefore, there is no scope for expressivity. Therefore, such strains with variable penetrance and expressivity can be polymorphic for that trait in terms of phenotype, even though all individuals have the same genotype.

In this part of the series, 28 different mutant phenotypes have been described. The concerned genes of these mutations are located on several different chromosomes of *D. melanogaster* (*Box* 4). In subsequent parts of the series, we will study a few aspects of transmission genetics with the help of these interesting mutants of *D. melanogaster* in order to take a closer look at patterns of inheritance of characters from parents to offspring.

## Suggested Reading

[1] H A Ranganath, Teaching and Learning Genetics with *Drosophila*, 1. *Drosophila* as a model system, *Resonance* 4(2), 48–52, 1999.

[2] D L Lindsley and G G Zimm, *The Genome of Drosophila melanogaster*, Academic Press, San Diego, USA, 1992.