Lecture 16—gene mutations

I. Gene mutations
   A. Change from wild type
      1. forward mutation
      2. reverse mutation (reversion)
      3. mutation

   B. Types mutations
      1. base-pair substitutions
         a. at DNA level
            i. transition
            \[ T \rightarrow C, C \rightarrow T, A \rightarrow G, G \rightarrow A \]
            ii. transversion
            \[ T \rightarrow A, T \rightarrow G, C \rightarrow A, C \rightarrow G \]
            \[ A \rightarrow T, G \rightarrow T, A \rightarrow C, G \rightarrow C \]
         b. at protein level
            i. silent mutation
            ii. conservative mutation
            iii. missense mutation
            iv. nonsense mutation

      2. frameshift

   C. Molecular basis of mutation

      - examples
II. Somatic vs germ line
A. somatic mutation

1. if in dividing cells
   - recessive
   - dominant

   - the earlier in development the mutation arises, the larger the mutant sector will be
2. if in non-dividing cells
   - most likely to have little or no effect
   - an exception is cancerous mutation
3. can somatic mutation be passed to progeny?
   - note that in plants germ line produced by somatic cells

B. germ line mutation

III. Mutant types
A. loss-of-function –
   null = no gene function

B. gain-of-function - generally dominant

C. 3rd type of dominant mutation: dominant negative

IV. Are mutations induced by selective conditions?
A. mutations are relatively random events
   - some mutations adaptive – permit growth under selective conditions (e.g. phage resistance)

B. pre-existing mutations are selected for
   - several experiments showed that mutations already present in population

   How was this demonstrated?
   - Replica plate (lederberg)

C. spontaneous mutation rates
   1. *E. coli*
   2. *Drosophila yellow*
   3. human