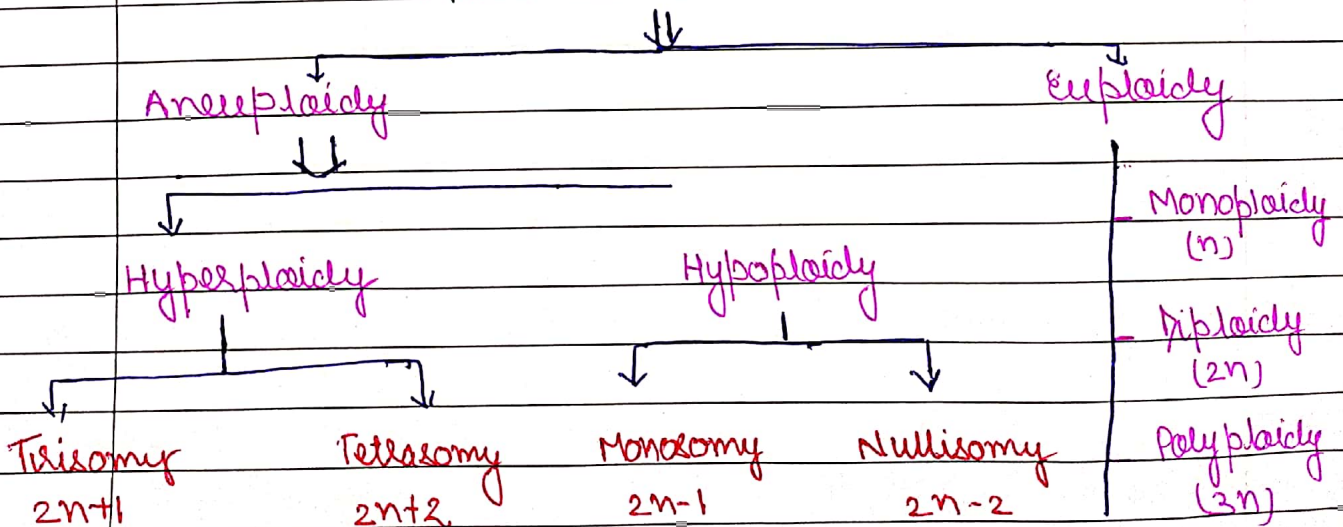


### Variation in chromosome number (Unit-II)

Part-2. Previous one is variation in structure

- \* Human cells normally contain 23 pairs of chromosomes, for a total of 46 chromosomes in each cell.
- \* A change in the number of chromosomes can cause problems with growth, development and function of the body's systems.
- \* Changes in number of whole chromosomes is called **aneuploidy**.
- \* Aneuploidy may involve entire set of chromosomes (**euploidy**), or loss or addition of single whole chromosome (**aneuploidy**).
- \* Each may produce phenotypic changes, modifications of phenotypic ratio or alteration of linkage group.

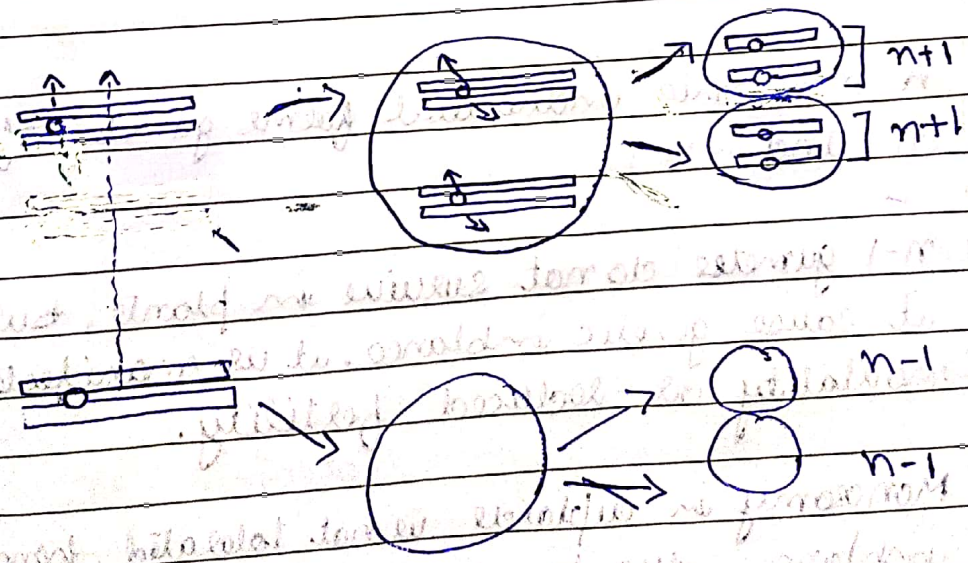
### NUMERICAL ABERRATION





# Aneuploidy

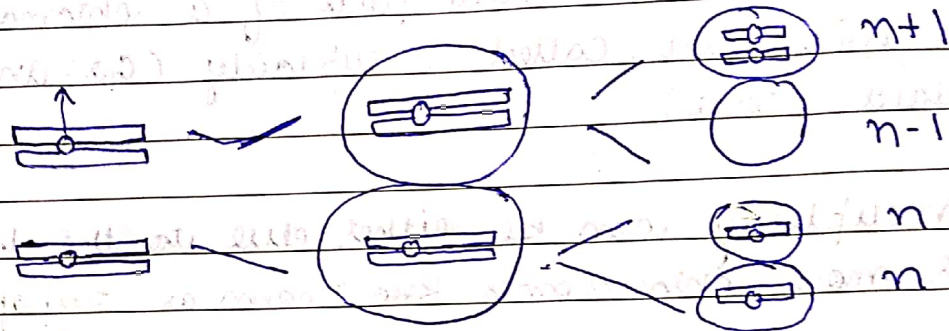
- \* changes that involve parts of a chromosome set results in individual, called aneuploidy (Gr. aneu = uneven; ploid = unit)
- \* Aneuploidy can be either due to the loss of one or more chromosomes known as hypoploidy
- \* Addition of one or more chromosome to the complete chromosome set known as hyperploidy.
- \* Hyploidy is mainly due to the subtraction of a single chromosome called monosomy ( $2n-1$ ) or due to the loss of one pair of chromosome called nullisomy ( $2n-2$ )
- \* Hyperploidy may involve addition of either a single chromosome called trisomy ( $2n+1$ ) or a pair of chromosome called tetrasomy ( $2n+2$ ).



Non-disjunction at first division



### Non-disjunction at second division



- ★ The mode of origin of a neuploid gametes by non-disjunction at either the first or second meiotic division

### ↓ Monosomy

- ★ Diploid organisms with all missing one chromosome of a single pair are monosomic with genomic formula  $2n-1$ .

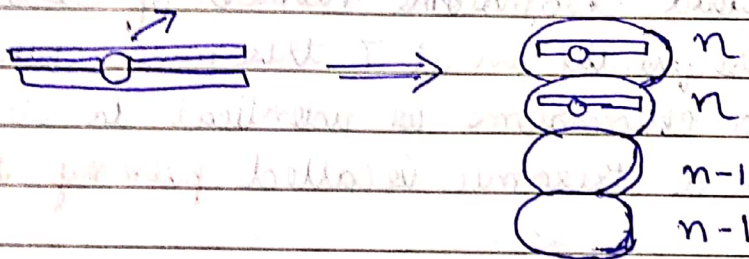
- ★ A monosomic individual forms gametes of two types, (n) and (n-1).

- ★ n-1 gametes do not survive in plants, but in animals it cause genetic imbalance, it is manifested by high mortality or reduced fertility.

- ★ Monosomy in diploids is not tolerated, hence it create imbalance due to loss of one complete chromosome.



- A The number of possible monosomics in an organism will be equal to the haploid chromosome number.  
for eg: in common wheat, since 21 pairs of chromosomes are present, 21 possible monosomics are known. Those 21 monosomics were produced by E-R seeds in variety called Chines spring.
- A Monosomics were also reported in cotton ( $2n=52$ ) by J. Endrizzi.



### Behaviour of a chromosome at meiosis

#### 2. Nullisomy

- A An organism which has lost a chromosome pair is a nullisomic.
- A The nullisomic organism has the genomic formula  $(2n-2)$ .
- A A nullisomic diploid often does not survive, a nullisomic polyploid (eg. hexaploid wheat,  $6n-2$ ) may survive but reduced vigour & fertility.



3. **Trisomy** :- It was first observed in batata elemanium by A.F. Blakeslee and J. Belling (1924).

+ Trisomics are those diploid organisms which have an extra chromosome ( $2n+1$ ).

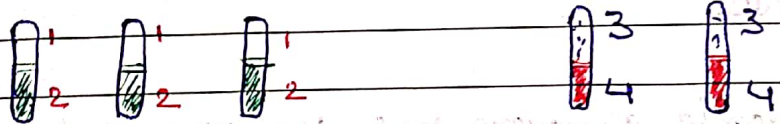
+ The extra chromosome may belong to any one of different chromosomes of a haploid complement, the number of possible trisomics will be equal to the haploid chromosome number.

For eg:- Haploid chromosome number of barley is 7, consequently in it 7 trisomics. When the extra chromosome is identical to its homologs such a trisomic is called **primary trisomics**.

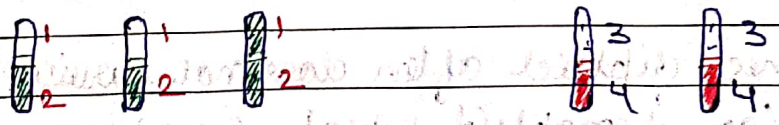
- When the extra chromosome should be an isochromosome (i.e. both chromosome arm genetically similar) is called **secondary trisomics**.

- When the extra chromosome should be the product of translocation.

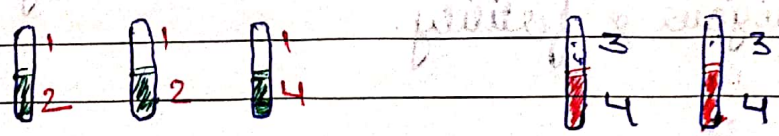
**Primary trisomic**



**Secondary trisomic**



**Tertiary trisomic**





## \* Trisomy in human

### 1) Down's Syndrome (DS) or Trisomy-21

\* Down's syndrome is named after the physician J. Langdon Down in 1866 it was formally called mongolism or mongolian idiocy.

\* It is usually associated with a trisomic condition for one of the smallest human autosome i.e. chromosome 21.

\* These include mild or moderate mental retardation, eyes that slant up & out; a tongue that is large, swollen & protruding; small and under developed ears; short stature; stubby fingers; an enlarged liver & spleen.

### 2. Edward Syndrome or Trisomy-18

\* It was first described in 1960 by John H. Edwards & his colleagues.

\* It is characterized by multiple malformations, primarily low-set ears; small receding lower jaw, flexed & clenched fingers; cardiac malformations & various deformities of skull, face & feet.



### 3. Patau syndrome or Trisomy-13

→ It is described in 1960 by ~~K~~ Klaus Patau

→ Symptoms appear to be mentally retarded; have sloping forehead, harelip & cleft palate

### 4. Double Trisomy

→ When two different chromosomes are represented in triplicate & double trisomics is resulted its genomic formula  $2n+1+1$

### 5. Tetrasomy

→ When the diploid organisms have two extra chromosomes are known as tetrasomic. They have genomic formula  $2n+2$ .



## 2. Euploidy

- \* The term euploidy (Gr. eu = even or true; ploid = unit) designates genomes containing chromosomes that are multiples of some basic number ( $x$ ).
- \* Euploids are those organisms which contain balance set or sets of chromosomes in any number.
- \* The number of chromosomes in a basic set is called monoploid number. When the number of set is greater than two called polyploid.
- \* Thus  $x$  is monoploid,  $2x$  is diploid and polyploid types are  $3x$  triploid,  $4x$  tetraploid,  $5x$  pentaploid,  $6x$  hexaploid.

### 1.) Monoploidy

- \* Monoploidy have a single set of chromosome.  
eg: 7 in barley and 10 in corn
- \* Monoploidy is common in plant but rare in animals.

### 1.) Origin & production of monoploids

- \* Monoploids are found normally & are produced due to parthenogenesis, as in male (drone) hymenopteran insects such as bees, wasps & ants.



\* In angiosperms monoplasts may also originate spontaneously due to parthenogenetic development of egg.

\* Such rare monoplasts have been obtained in tomato & cotton under cultivation.

\* Monoplasts can be produced by artificial means by following method

- 1) X-ray treatments
- 2) Delayed pollination
- 3) Temperature shock
- 4) Colchicine treatment
- 5) Distant hybridization
- 6) Anther or pollen culture

\* Among these, distant hybridization & anther culture

## 2) Morphology of monoplasts

\* Monoplast plants have reduced size of all vegetative & floral parts.

\* In monoplast *Nicotiana* reported that the leaves, flowers and overall plant size were smaller.

\* The size of seeds & stomata as well as diameter of pollen were found smaller in monoplasts than in the diploids.



### 3) uses of monoplastoids

A In a monoplastoid, since there is only one copy of each chromosome, only one allele of each gene, so in it each gene is expressed, whether it is dominant or recessive.

A Success has been achieved in developing monoplastoid strain Nicotiana, Datura and Triticum.

### 2) Polyploidy

A Any organism with more than two genomes ( $2n$ ) is called a polyploidy.

A For eg: - the rose genus Rosa include species with the somatic number 14, 21, 28, 35, 42 & 56. These number all the multiple of 7. This is a euploid series of the basic monoplastoid number 7 which gives ~~2n~~  $2n$ ,  $3n$ ,  $4n$ ,  $5n$ ,  $6n$  and  $8n$  species.

### Types of Polyploids

- 1) Autopolyploids
- 2) Allopolyploids
- 3) Autallopolyploids







## i) Origin & production of autopolyploids

A The autopolyploids may occur in nature or produced artificially.

A ~~newer~~ one of the common examples of natural autopolyploidy is "doob grass" (Cynodon dactylon), where its autopolyploidy nature is deduced by their meiotic behavior.

↳ Polyploidy may arise by:-

i) as a result of interference with cytokinesis, once chromosome replication has occurred.

ii) It may occur in somatic tissues which give rise to tetraploid branches.

A Some examples of artificial methods are seedless varieties of watermelon, sugar beet, tomato, grapes & banana.

A Autopolyploidy have been induced by artificial means such as chemical (colchicine, mercury chloride, chloral hydrate), radioactive substance (radium & X-rays) and temperature shock.

A ~~These~~ ~~induces~~ ~~usually~~ ~~disturb~~ the mitotic or meiotic spindle formation and ~~may~~ cause non-segregation of already duplicate chromosomes, during cell division. #

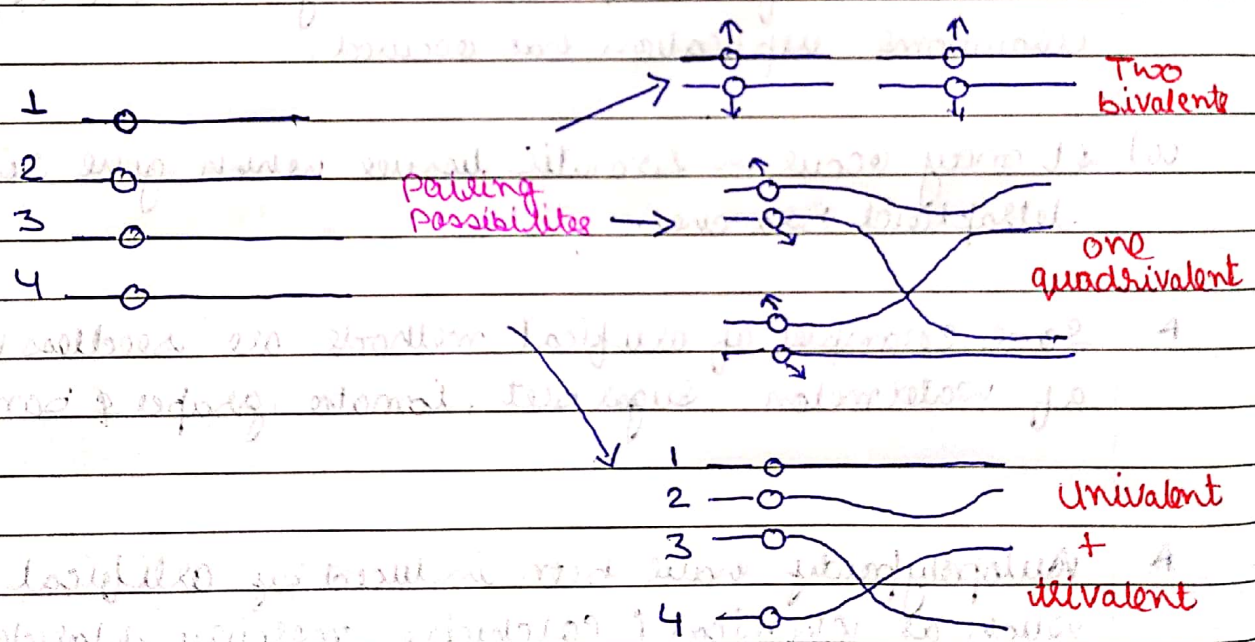


(ii) Effects of autopolyploids.

→ It results in gigantism of plant cells i.e. leaves, flowers & fruits of an autopolyploid are larger in size than a diploid plant.

→ For eg:- the size of lower epidermis of leaf of a tetraploid Saxifraga pennsylvanica was found greater than diploids

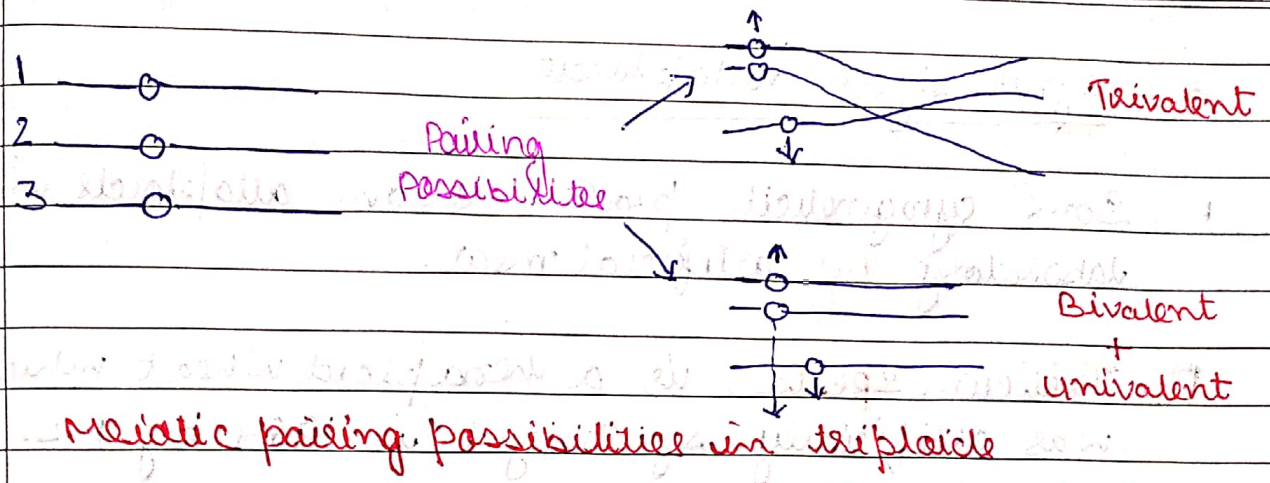
\* Polyploid varieties with an even number of genomes (eg tetraploids) are often fully fertile, whereas



Meiotic pairing possibilities in tetraploids

→ whose variety have an odd number (eg triploids) are highly sterile



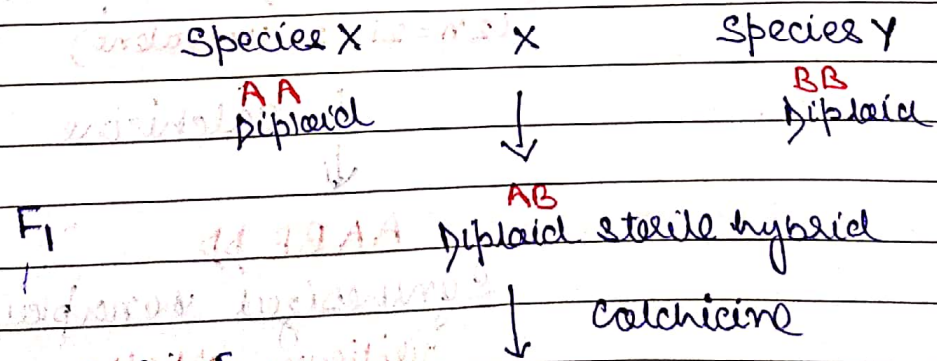


Meiotic pairing possibilities in triploid

ii) Allopolyploids

When the polyploidy results due to the doubling of chromosome number in a  $F_1$  hybrid which is derived from two distinctly different species, then, it is called allopolyploidy.

Let, A represent a set of chromosome in species X and B represent another genome of species Y. The  $F_1$  hybrid of these species will have one A & another B genome.



Doubling of chromosome in  $F_1$  hybrids will give allotetraploids with 2 A & 2 B genome

**AARR**

Amphidiploid tetraploid Fertile.

classmate Formation of an amphidiploid tetraploid



\* Synthesized allopoloids

Some cytogeneticists produce certain allopoloids in laboratory by artificial mean.

\*) Triticum spelta, is a hexaploid wheat which was artificially synthesized in 1964 by F.S. McFadden & E.R. Sears.

\* They synthesized crossed of Triticum dicoccoides (tetraploid  $2n=28$ ) with goat grass Aegilops squarrosa (diploid  $2n=14$ ) & doubled the chromosome number in  $F_1$  hybrid.

Triticum dicoccoides X Aegilops squarrosa

AA BB

DD

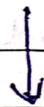
( $2n=28$ ; 14 bivalent)

( $2n=14$ ; 7 bivalent)



ABD - Triploid hybrid

( $2n=21$ ; 21 univalent)



calchicine

AA BB DD

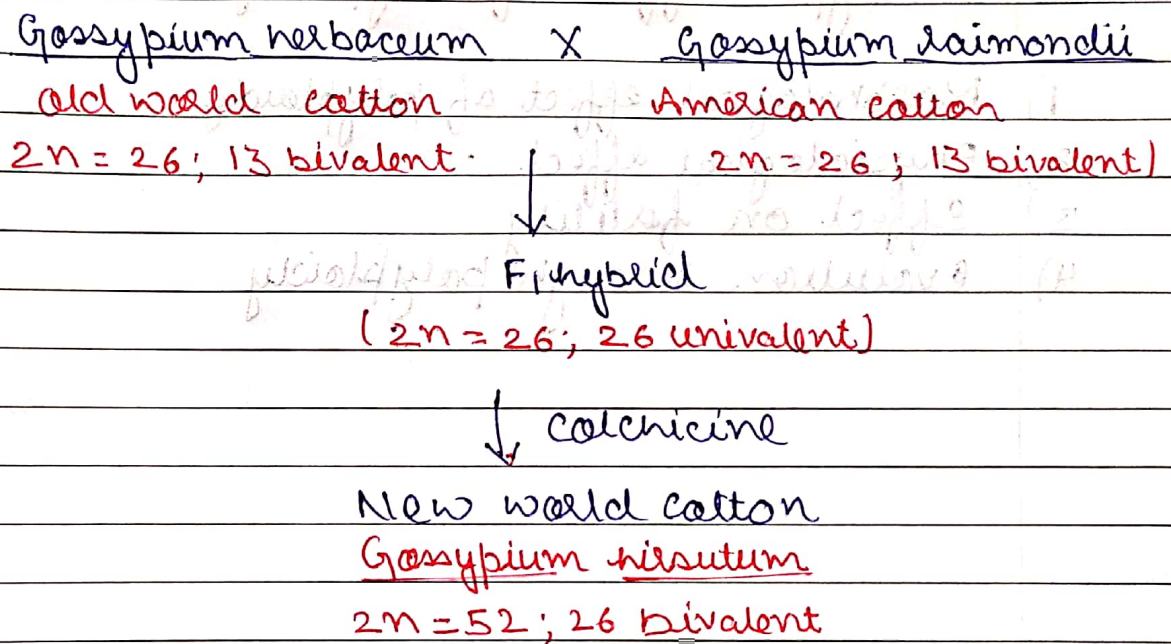
Synthesized hexaploid wheat

Triticum spelta

( $2n=42$ ; 21 bivalent)



(ii) Gossypium hirsutum, the new world cotton plant is the another example of it.  
J.O Beasley crossed the old world & American cotton and doubled the chromosome number in the  $F_1$  hybrids



### 3) Autoallopolidy

▶ In this one genome is in more than diploid state. Commonly autoallopolidy are hexaploids. (AAAABB) eg:- Helianthus tuberosus.

▶ This term was used by Koestoff in 1939.

▶ Polyploidy was found rare in animals but occur in flatworms, leeches and brine shrimp.