Incidence:

Down syndrome occurs in approximately 1 in 600 births.

Male: female ratio at birth is 1.24 : 1.0

Genotypes of Down syndrome:

1-Meiotic non-disjunction trisomy 21 95%

2 - Translocation Trisomy 4%

3- Mosaic type 1%

1. Meiotic non-disjunction (Maternal age dependent DS)

- All the cells show an additional No. 21 chromosome, i.e. 47, XX, +21. This type is resulting from meiotic non-disjunction.

- Approximately 95% of all Down syndrome are of this type.

•About 70% of trisomy 21 is born to mothers over 30 years.

•However, karyotyping of both parents is usually normal.

2 - Translocation Trisomy 21

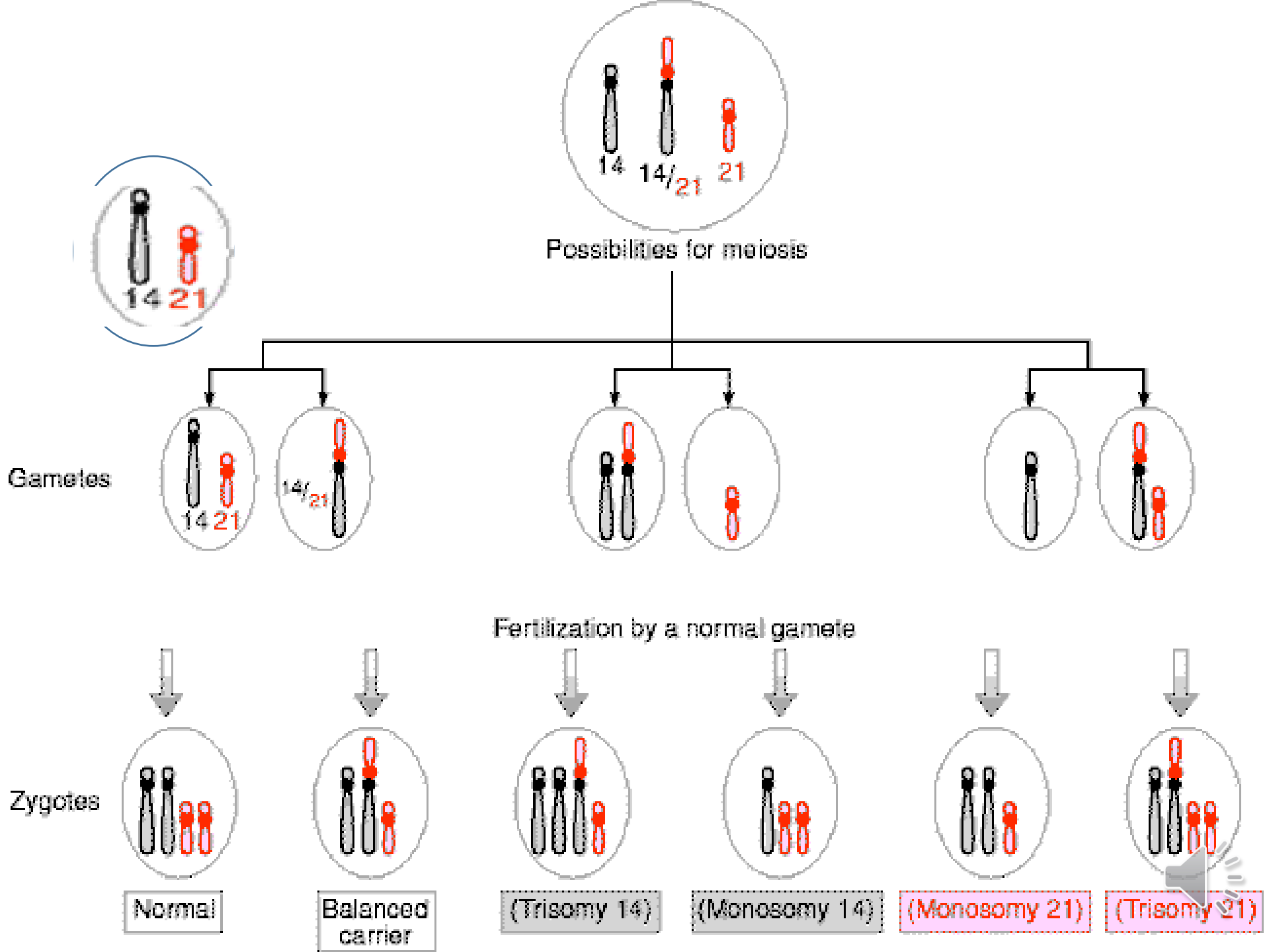
•All cells show normal number of chromosomes (46), however an extra-chromosome 21 is attached to another one (13,14, 15 or 21) i.e. 46, XX, +t (14q 21q).

•Approximately 4% of Down syndrome is of this type.

The mechanisms of underlying translocation are:

•**Sporadic type**: half of translocated Down syndrome arise de novo in the affected individual. Both parents are usually normal.

•**Inherited type**: another half inherited the disease from a balanced translocation carrier parents i.e. 45, XX, t (14q 21q).


14
14/21
21
Possibilities for meiosis
14 21
Gametes
14 21
14/21
Fertilization by a normal gamete
Zygotes
Normal
Balanced carrier
(Trisomy 14)
(Monosomy 14)
(Monosomy 21)
(Trisomy 21)

3. **Mosiacism:** (Mitotic non-disjunction).

- This type represents 1% of Down syndrome.

- They have various proportions of trisomy 21 (47, XX, + 21) and normal cells (46, XX).

- This group may be of normal intelligence, depending on the number of trisomic cells present.
- . The parents are usually normal