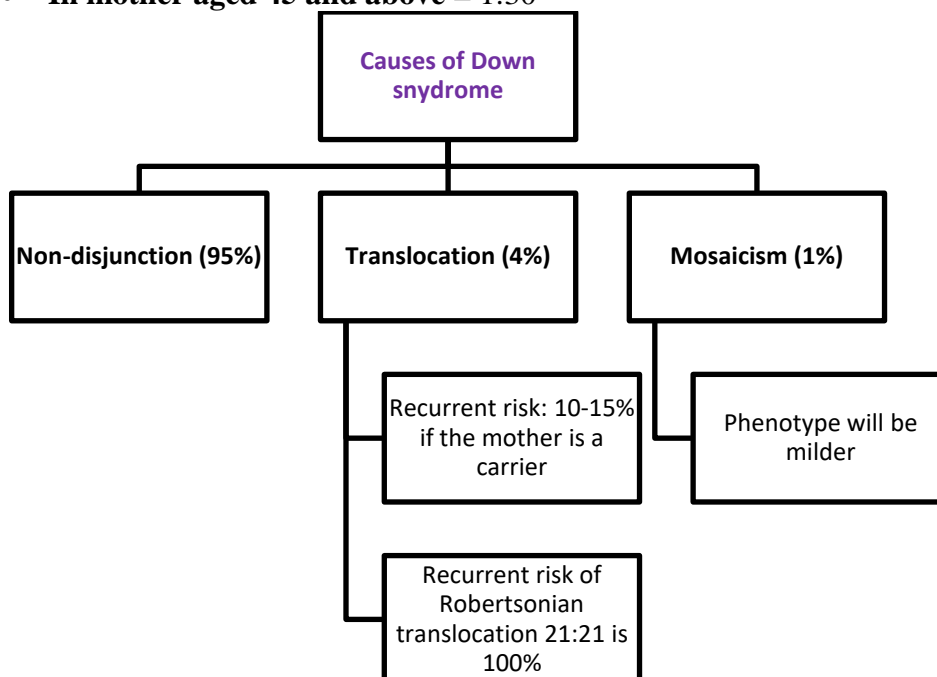




Incidence:

- **Worldwide** = 1:600-1:800
- **In mother aged 45 and above** = 1:30



Clinical features of Down syndrome:

Head and neck	Blood
<ol style="list-style-type: none"> 1. Round face and flat nasal bridge 2. Epicanthal skin folds 3. Short neck and excessive nuchal skin 4. Upward slanting eyes 5. Cataracts and glaucoma 	<ol style="list-style-type: none"> 1. Increased risk of leukemia
Limbs	CNS
<ol style="list-style-type: none"> 1. Short 5th finger of the hand 2. Single palmar crease 3. Wide space between 1st and 2nd toes 	<ol style="list-style-type: none"> 1. Hypotonia 2. Developmental delay 3. Alzheimer's disease
Heart	Skin
<ol style="list-style-type: none"> 1. Arterial Septal Defect (ASD) 2. Ventricular Septal Defect (VSD) 3. AV septal defect 4. Patent Ductus Arteriosus (PDA) 	<ol style="list-style-type: none"> 1. Fissured tongue
GI	
<ol style="list-style-type: none"> 1. Duodenal atresia 2. Imperforate anus 	
Endocrine	
<ol style="list-style-type: none"> 1. Hypothyroidism 2. Male: 100% infertile 3. Females can get pregnant 4. Obesity and short stature 	

Diagnosis:

- **Antenatal:** amniocentesis, CVS or triple test.
- **Postnatal:** by phenotype and this will be confirmed through karyotyping and FISH