Malformation

This article contains probably doubtful information.

The article "Malformation" contains probably doubtful information. More detail information can be found on its talk page.

Malformations are congenital developmental defects – congenital morphological deviations exceeding normal variability associated with significant form and functional disorders.

Normal intrauterine and postnatal development depends on the gradual activation or suppression of genes, the zygote contains all genes, but most of them are inactive, with the gradual development of the fertilized egg, individual genes or their groups are involved.

Basic Concepts

anomaly - a small deviation that does not lead to a functional malfunction, an even smaller degree of deviation is called **variation**

agenesis - complete absence of an organ and its foundations, or the absence of some cell population in the tissue

aplasia - absence of an organ, but with preservation of its base or rudiment of structure (e.g. lung aplasia - the main bronchus is blindly terminated in rudimentary lung tissue)

hypoplasia - congenital reduction of all components of an organ (microphthalmia, microcephaly, micrognathia, etc.), in contrast to atrophy, which is an acquired reduction of a normally developed organ

atresia - failure to form the lumen of a hollow organ (some organs (e.g. esophagus) are formed as solid strips, the central cells of which die by apoptosis and thus a cavity is formed)

dysplasia - abnormal arrangement of cells in tissues or the presence of tissues that do not normally occur in a given organ (must be distinguished from dysplasias in the sense of precancerous changes - intraepithelial neoplasias)

ectopy (heterotopia) - abnormal placement of an organ (e.g. ectopia of the heart - heart outside the chest cavity)

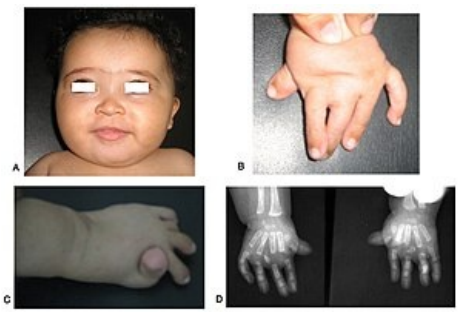
dystopia - the organ remains in the place of its original development (e.g. kidney dystopia - kidney in the pelvis)

failure of involution - persistence of certain embryonic structures that normally disappear (e.g. persistence of ductus thyreoglossus)

separation disorders - failure of the apoptosis mechanism, e.g. syndactyly

dysraphia - incomplete closure (e.g. spina bifida and other "split" defects)

Causes



Temtam preaxial brachydactyly syndrome

The causes are referred to as **teratogens** and can be:

- internal (genetic influences – structurally abnormal genes or gene activation disorder);
- external (on the part of the mother – exposure of the fetus to physical and chemical influences), the result of the action of the teratogenic agent depends on:
  - specific effect (e.g. blocks cell division, damages cellular respiration...);
  - stage of development in which the given organs are sensitive to the action of the teratogen.

For individual teratogens, there are periods during the development of the embryo when they can be used (so-called *critical periods*) and when they are not, the critical periods for different parts of the organism can be the same - the effect of one agent then creates a combination of defects (syndrome). The same congenital defect can be caused by different influences, and conversely, one factor can cause different defects.

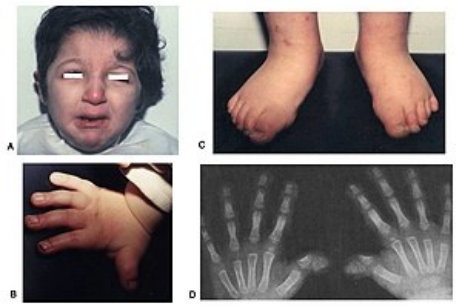
1. **physical teratogens** - radiation
2. **chemical teratogens** - drugs (transplacental transfer - thalidomide, cytostatics, antiepileptics, retinoids)
3. **biological teratogens** - transplacental transfer of infection (toxoplasmosis, rubella, CMV, HSV)
4. **other suspected teratogens** - smoking, alcoholism, diabetes and maternal circulatory disorders

Multiple malformations can arise as a *polyotropic effect* of the teratogen (several organs are affected at the same time) or as a *monotropic effect* (injury of one organ causes a cascade of pathogenetic events, e.g. kidney agenesis - oligohydramnios - deformation of the fetus due to uterine pressure - lung hypoplasia) .

Developmental syndrome - the only cause for anomalies in distant organs (polytropic teratogen effect).
Developmental association (syntropy) - a statistically significantly more frequent association of changes, no single cause is known.

Division

From the point of view of the period when the developing individual was damaged, different types of malformations are distinguished:



Rubinstein-Taybi syndrome

1. **gametopathy** - gamete damage - most often chromosomal aberration;
2. **blastopathy** - disorders of the fertilized egg (blastocyst) up to the 15th day of development - usually very severe defects incompatible with life, rare conjoined twins, teratomas;
3. **embryopathy** - disorders of embryo development from the 15th day to the end of the 3rd month of pregnancy - during this period, organogenesis takes place (all organs are formed), damage at the beginning of organogenesis leads to very severe defects, at the end, milder defects occur, it is, for example, cleft defects of the spine and spinal cord or the palate and face;
4. **fetopathy** - disorders of the fetus from the end of the 3rd month of pregnancy until birth, the basic development of the organ is complete, its components continue to develop, examples are disorders of the development of certain groups of neurons in the brain or the vascular supply of the myocardium.

Blastopathy

Monster duplex (Siamese twins)

– incomplete separation.

and. symmetrical (both monsters the same)

- *thoracopagus* (sternopagus, xiphopagus) – thoracic joint
- *pygopagus* (ischiopagus) – pelvic joint
- *craniopagus* – head joint
- *prosopo-cephalo-thoracopagus* (Janus) - fusion of heads and chests with faces facing the shoulders
- *dicephalus* - parallel fusion of trunks, two spines, usually only two pairs of limbs
- *dipygus* – parallel fusion of heads and chests from the side, can have two HK and four DK

b. asymmetrical (one monster larger – autosit, the other smaller or rudimentary – parasit)

- *epignathus* – a parasite anchored at the base of the skull, protruding from the mouth of the autosita
- *parasitic thoracopagus*
- *parasitic pygopagus* (more differentiated than sacral teratoma, e.g. with a hint of a limb)
- *fetus in feto* – an internal monster, e.g. in the abdominal cavity

Monster simplex

Grossly externally deformed individual (e.g. anencephalus) or internal disorders (situs inversus seu heterotaxis).

Embryopathy

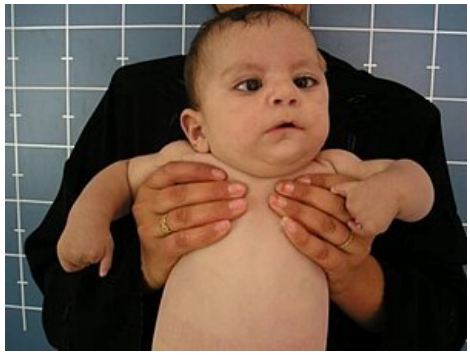
Cleft defects of the spine and spinal cord

These are not true clefts, but disorders of the medullary disc closure (dysraphia), manifested as cranioschisis, rhachischisis or craniorhachischis.

a. cranioschisis (split head), can be partial (*hemicrania* - preserved rudiments of the vault bones) or complete (*acrania* - completely missing the skull cap), acrania is combined with anencephaly, when the brain is missing - a malformation incompatible with life (in survivors, the trunk is preserved), failure of the pituitary gland leads to underdevelopment of the adrenal glands, only the facial part and base are developed from the skull, the forehead bends sharply backwards and passes into the base, which is not covered by skin, but by vascular tissue with rudiments of nerves tissues (*area neurovasculosa*), the bulbs are well formed and strongly protrude, the eyes look upwards (*uranoscopy*), in case of *craniorhachischis* it resembles a toad monster (the neck is missing and sits directly on the trunk, uranoscopy, area neurovasculosa smoothly transitions into an analogous structure on the dorsal side of the body)

> **encephalocele** - a defect in the neurocranium associated with a protrusion of the brain and its coverings, it takes the form of a cystic spherical mass most often in the occipital area covered with skin, the contents are the hard membrane (meningocele), sometimes the brain (encephalocele)

b. rhachischisis - a defect in the closure of the spinal canal (splitting of the vertebrae), it can be total (often associated with cranioschisis) or partial (most often in the lumbar region), depending on the place where the bases of the spine are not joined, we can distinguish *rhachischisis anterior, lateralis et posterior*, the most significant is *rhachischisis posterior* - if the skin cover does not develop at all (*myeloschisis*, a defect incompatible with life), there is a strip of rudimentary nerve tissue (*area medullovasculosa*) in the middle of the back, which passes along the edges in the skin, the rudiments of the spinal roots enter it, the skin on the edges is mostly overgrown with hair (*hypertrichosis*), defects with a preserved skin cover belong to the group of *spina bifida (cystica et occulta)*, Complications of rachischis are innervation disorder in the cauda equina area and the possibility of ascending infection of the diaper and brain


Möbius syndrome

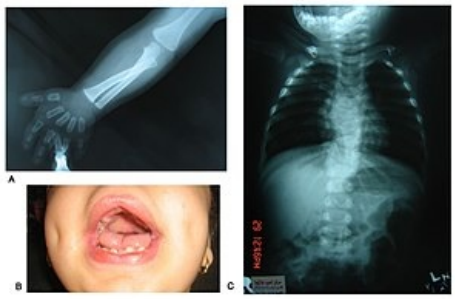
spina bifida cystica - further divided into:

- *meningocele* – the dura and arachnoid form a sac arched into the skin, the spinal cord has a normal position or runs through the center of the sac, the contents of the sac are cerebrospinal fluid;
- *meningomyelocele* – the spinal cord is located in a bag and connected to its surface ectoderm;
- *hydromyelomeningocele* – a similar defect to the previous one, but the spinal cord has a dilated central canal (hydromyelia);
- *myelocystocele* - the spinal cord forms an area medullovasculosa, which passes peripherally into the skin, fluid accumulates ventrally, arching the entire structure dorsally;
- *spina bifida occulta* - dorsal split of the spine, which is covered by a stiff fibrous membrane above the spinal cord, there is no arching, the skin over the defect is thickened, sometimes with hypertrichosis.

Cleft facial defects

They include isolated or combined defects (e.g. Pierre-Robin syndrome = macroglossia + micro-gnathia + palatoschisis).

- *cheiloschisis* - cleft lip
- *gnathoschisis* - cleft jaw
- *palatoschisis* - cleft palate
- *uranoschisis* - cleft of the uvula


Robin syndrome

Fetopathy

They include deformations of normal organ bases by internal or external mechanical force (distortions of shape, displacement of organs by tension or pressure):

- **compression** of the fetus from the anomaly of the uterus;
- **lack of movement** in oligohydramnios, myopathies, neurological disorders;
- **detachment**, interruption of the normal basis of an organ with vascular damage;
- amniotic **adhesions** and streaks leading to limb strangulation;
- **vascular damage**, e.g. abuse of cocaine or amphetamines in the mother will cause vascular disorders in the fetus.

The same malformation can arise by different mechanisms, e.g. pes equinovarus is the result of insufficient movement of the limb from the pressure of the uterine wall and from a spinal cord disorder.

Links

Source

PASTOR, Jan. *Langenbeck's medical web page* [online]. ©2006. [feeling. 2009-01-17]. <https://langenbeck.webs.com/>.