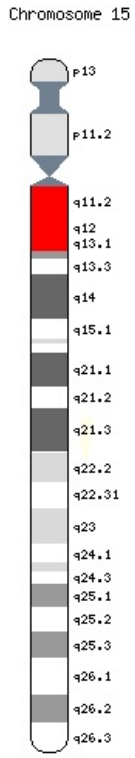
# Microdeletion syndromes

**Microdeletion syndromes** are a specific group of structural chromosomal aberrations caused by **deletions** of a very small scale (hence the name of this group of syndromes). We classify these small-scale deletions among the so-called *submicroscopic chromosomal aberrations* - because during a routine karyotype examination in an optical microscope they can no longer be detected by the human eye (the theoretical limit is approx. 5 million bases [1]). These syndromes are also called contiguous gene syndromes or autosomal segmental aneusomy syndromes*, as their clinical symptoms are caused by a failure of expression* more gens. In general, these syndromes have very diverse clinical manifestations, but practically all of them have some degree of psychomotor retardation and craniofacial dysmorphia.

These small interstitial deletions arise by **unequal crossing over** between homologous chromosomes or unequal exchange between sister chromatids. This group of diseases also includes some syndromes associated with **imprinting disorder,** where the clinical manifestations of the deletion differ depending on whether the chromosome of maternal or paternal origin is damaged (Prader-Willi syndrome, Angelman syndrome).

When diagnosing these syndromes, it is usually not enough to carry out a **routine karyotype examination**, but more detailed methods must be used. When a specific syndrome is clinically suspected, it is possible to use a locus-specific **FISH** probe, the disadvantage is the very limited scope of such an examination (a special probe is required for each tested locus/syndrome), therefore in practice not entirely clear clinical diagnoses use rather methods that are able to test a larger number of syndromes at the same time - for example **MLPA**, or nowadays increasingly often **microarray**.



Chromosome 15 - red: deletion in Angelman syndrome

## Selected examples of microdeletion syndromes

**Microdeletion syndromes – autosomal segmental aneusomy syndromes**

| Abbreviation | Syndrome | Deletion area |
|---|---|---|
| WAGR | Wilms tumor, aniridia, urogenital anomaly, MR | del 11p13 |
| RB | Retinoblastoma | del 13q14 |
| PWS | Prader-Willi syndrome | del 15q11-13 |
| AS | Angelman Syndrome | del 15q11-13 |
| MDS | Miller-Dieker syndrome-lissencephaly | del 17p13 |
| DCS/VCFS | Di George/velocardiofacial syndrome + conotruncal heart defects | del 22q11 |
| LGS | Langer-Giedeon syndrome (tricho-rhino-phalangeal syndrome) | del 8q24.11-24.13 |
| SMS | Smith-Magenis syndrome | del 17p11.2 |
| WS | Williams syndrome | del 7q11.23 |
|  | Cri du chat syndrome | section 5p15.2 |

# Links

## Related Articles

- Chromosomal Abnormalities
- Structural Chromosomal Aberration
- Gene imprinting and human pathologies
- Indications for karyotype examination

## References


1. EMANUEL, Beverly S – SAITTA, Sulagna C. From microscopes to microarrays: dissecting recurrent chromosomal rearrangements. *Nat Rev Genet* [online]. 2007, vol. 8, no. 11, p. 869-83, Available from <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2858421/?tool=pubmed>. ISSN 1471-0056 (print), 1471-0064.