

MONOSOMY

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MONOSOMY

Primary Disciplinary Field(s): Genetics, Cytogenetics, Cell Biology

1. Core Definition

Monosomy is a specialized form of aneuploidy characterized by the presence of only one copy of a particular chromosome in an otherwise diploid cell ($2n-1$), rather than the typical two copies present in a healthy organism. This genetic condition results from a severe error during cell division, specifically **meiosis**, where one member of a homologous pair of chromosomes is absent in the resulting gamete. When this deficient gamete fuses with a normal gamete, the resulting zygote lacks one full chromosome, leading to a profound **gene dosage imbalance**. Such an imbalance means that genes normally requiring two copies for proper regulation and expression are present in only a single copy, often leading to insufficient protein production (haploinsufficiency) or the detrimental expression of recessive lethal genes.

The concept of monosomy is fundamentally tied to the larger category of **aneuploidy**, which describes any deviation from the euploid (normal) number of chromosomes. While aneuploidy encompasses both the gain (trisomy, $2n+1$) and loss (monosomy, $2n-1$) of whole chromosomes, monosomy tends to be far more damaging to human development than trisomy. The complete lack of an entire set of genes typically proves non-viable, leading to high rates of spontaneous abortion, particularly when involving large **autosomal chromosomes**. Therefore, clinically viable forms of full monosomy are exceedingly rare, primarily limited to monosomy of the sex chromosomes.

It is crucial to differentiate between full monosomy, where an entire chromosome is missing, and **partial monosomy**, where only a segment of a chromosome is deleted. Partial monosomies can be survivable, though they invariably result in significant developmental syndromes, such as Cri-du-chat syndrome (deletion on chromosome 5). However, when the term monosomy is used without qualification in a clinical or academic setting, it refers to the absence of one entire chromosome, a condition that disrupts hundreds or thousands of crucial genetic interactions necessary for successful embryogenesis.

2. Etymology and Historical Development

The term **Monosomy** is derived from Greek roots: 'mono' meaning **single** or **one**, and 'soma' meaning **body**, referring to the chromosome body. The theoretical basis for understanding numerical chromosomal abnormalities emerged in the early 20th century following the rediscovery of Mendel's laws and the establishment of the **chromosome theory of inheritance**. Early geneticists recognized that deviations from the normal chromosomal complement could explain specific inheritance patterns and developmental defects observed in organisms like the fruit fly,

Drosophila melanogaster.

The systematic investigation into human chromosomal disorders began in earnest in the late 1950s. The breakthrough came in 1956 when Joe Hin Tjio and Albert Levan correctly determined the human diploid chromosome number to be 46. This accurate count paved the way for identifying specific aneuploidies. Prior to this, accurate **karyotyping** techniques were insufficiently developed. Shortly thereafter, in 1959, the most common human trisomy, Down Syndrome (Trisomy 21), was identified by Jérôme Lejeune.

Following the identification of trisomies, researchers confirmed the existence of monosomies. The first, and most clinically significant, human full monosomy identified was **Monosomy X**, also known as Turner Syndrome, which was thoroughly characterized in the mid-20th century. The discovery of Turner Syndrome demonstrated that while autosomal monosomies were generally lethal, the reduced gene content of the X chromosome, coupled with the natural process of X-inactivation in females, allowed for conditional viability, solidifying the importance of chromosome size and functional redundancy in determining the severity of aneuploidy.

3. Key Characteristics

Haploinsufficiency: A primary characteristic of monosomy is **haploinsufficiency**, the mechanism by which a single functional copy of a gene is insufficient to produce the necessary gene product (protein or RNA) to ensure a normal phenotype. Since hundreds or thousands of genes are affected when an entire chromosome is missing, the resulting cellular and developmental defects are profound and often incompatible with life.

Lethality in Autosomes: Full monosomy involving any of the 22 pairs of **autosomal chromosomes** (non-sex chromosomes) is almost universally lethal in humans. The resultant zygote typically fails to implant or results in early spontaneous abortion. This high lethality rate underscores the necessity of precise gene dosage regulation across all major chromosomes for the complex orchestration of embryonic development.

Viability Restricted to Sex Chromosomes: The only known full monosomy compatible with human postnatal survival is **Monosomy X** (45, X), which causes Turner Syndrome. This is tolerable because the X chromosome is subject to **X-inactivation** (dosage compensation) in normal females (XX), meaning that one X chromosome is already silenced early in development. The absence of a second sex chromosome therefore results in less severe dosage disruption compared to the absence of an autosome.

Mosaicism: Monosomy often presents in a mosaic form, meaning only a fraction of the body's cells carry the $2n-1$ complement, while the rest are normal ($2n$). **Mosaic monosomy** is generally less severe than non-mosaic or uniform monosomy, as the presence of a healthy cell line can

compensate for the deficient cells, sometimes allowing for extended survival or reduced clinical severity.

4. Mechanism of Origin: Nondisjunction

The overwhelming cause of monosomy is **nondisjunction**, an error that occurs during the process of **meiosis**--the cell division necessary to produce gametes (sperm and egg). Nondisjunction refers to the failure of homologous chromosomes or sister chromatids to separate properly. If nondisjunction occurs, it leads to the production of abnormal gametes that either have an extra chromosome ($n+1$) or are missing a chromosome ($n-1$).

Nondisjunction can occur during **Meiosis I** when homologous chromosomes fail to separate. In this scenario, the resulting secondary spermatocytes or oocytes will either have both members of the homologous pair or neither. If the gamete resulting from this process is missing a chromosome, and it is fertilized by a normal gamete (n), the resulting zygote will be monosomic ($2n-1$). This type of error accounts for a significant portion of aneuploidies.

Alternatively, nondisjunction can happen during **Meiosis II**, where sister chromatids fail to separate. If the primary division (Meiosis I) was normal, but Meiosis II fails for a particular chromosome, the resulting gametes will include two normal gametes (n), one trisomic gamete ($n+1$), and one monosomic gamete ($n-1$). Regardless of whether the error occurs in Meiosis I or II, the end product is a germ cell with a missing chromosomal body, providing the genetic basis for monosomy upon fertilization. Advanced maternal age is a well-established risk factor that increases the likelihood of nondisjunction events.

5. Clinical Consequences and Examples

The clinical expression of monosomy is largely dependent upon which chromosome is affected and whether the condition is full or partial. For nearly all **autosomal monosomies**, the consequence is early embryonic mortality. These severe chromosomal abnormalities are a leading genetic cause of miscarriage, often before a pregnancy is even clinically recognized. The few reported cases of live-born individuals with full autosomal monosomy (e.g., Monosomy 21) are exceedingly rare and usually involve high degrees of **mosaicism** or are associated with complex translocations.

The most significant clinical example of a survivable full monosomy is **Turner Syndrome** (Monosomy X). Individuals with Turner Syndrome are phenotypically female and typically present with a karyotype of 45, X. Common clinical features include **short stature**, primary ovarian insufficiency (gonadal dysgenesis), characteristic neck webbing, skeletal abnormalities, and potential cardiovascular defects. Management often involves **growth hormone therapy** to address height limitations and estrogen replacement therapy to induce secondary sexual characteristics and mitigate long-term health risks associated with estrogen deficiency.

In addition to full monosomies, partial monosomies result in distinct syndromes. One well-known example is **Cri-du-chat syndrome**, caused by a terminal or interstitial deletion on the short arm of chromosome 5 (5p-). This partial monosomy leads to severe intellectual disability, microcephaly, and the characteristic high-pitched, cat-like cry that gives the syndrome its name. The study of partial monosomies helps researchers map specific genes to localized regions of chromosomes, revealing the precise genetic segments responsible for certain developmental traits or pathologies.

6. Significance and Impact

The study of monosomy offers profound insights into fundamental biological processes, primarily concerning **gene dosage effects** and the principles of developmental robustness. Monosomic states demonstrate the extreme sensitivity of the human genome to variations in gene number. The lethal nature of most autosomal monosomies highlights that normal human development requires two precisely regulated copies of almost every gene; removal of even one copy is generally catastrophic, disrupting critical metabolic and signaling pathways.

Furthermore, understanding monosomy is critical in the field of **reproductive medicine** and prenatal diagnosis. Knowledge of monosomy allows clinicians to accurately assess the risk factors associated with pregnancy, particularly in older mothers, and to interpret the results of prenatal screening technologies such as **Non-Invasive Prenatal Testing (NIPT)** and diagnostic procedures like amniocentesis and chorionic villus sampling (CVS). Identifying monosomies prenatally provides families with necessary information regarding the prognosis and potential challenges of the pregnancy.

Monosomy research also informs cancer biology. While germline monosomy is typically lethal, somatic cells can develop monosomy (loss of heterozygosity) in specific chromosomes, which is frequently implicated in tumor development. For instance, the loss of one copy of a chromosome containing a **tumor suppressor gene** can contribute to oncogenesis, illustrating that the principles of gene dosage imbalance apply equally to developmental and pathological cellular contexts.

7. Debates and Criticisms

Monosomy, particularly its association with severe developmental syndromes and high rates of miscarriage, sits at the center of ongoing ethical and clinical debates surrounding **prenatal screening** and selective termination. As diagnostic technologies become more sensitive, detecting complex aneuploidies, including low-level mosaic monosomies, becomes increasingly common, forcing difficult decisions regarding the viability and quality of life for the affected individual. Critics often raise concerns about the ethical implications of using advanced screening to identify conditions that are highly likely, but not guaranteed, to result in severe impairment.

A key clinical challenge involves the complexity of **mosaic monosomy**. Distinguishing between a

true, uniformly affected fetus and one with clinically insignificant low-level mosaicism--where the majority of cells are healthy--is often challenging. Diagnosis based solely on fetal cell-free DNA (NIPT) can be complicated by placental mosaicism, where the abnormality is confined to the placenta and not the fetus, leading to potential **false positive diagnoses** for monosomic conditions and unnecessary anxiety or intervention.

Furthermore, there is continuous research and debate regarding the management protocols for survivable monosomies like Turner Syndrome. Optimizing **hormone replacement therapy**, addressing neurological and cognitive deficits, and providing comprehensive long-term care requires continuous reassessment as scientific understanding of the full spectrum of gene dosage effects evolves. The high variability in clinical presentation, even among individuals with the same 45, X karyotype, necessitates individualized treatment strategies, which remains a subject of ongoing clinical investigation.

Further Reading

[Monosomy \(Wikipedia\)](#)

[Aneuploidy \(Wikipedia\)](#)

[Turner Syndrome \(Monosomy X\) \(Wikipedia\)](#)

[Nondisjunction \(Wikipedia\)](#)