

Chromosome 5

Chromosome 5 is one of the 23 pairs of human chromosomes and is classified as an autosome. Here are some key facts about it:

General Characteristics - **Size:** Chromosome 5 is one of the larger chromosomes, spanning approximately **181 million base pairs**. - **Gene Content:** It contains around **900-1,000** protein-coding genes. - **Location of Centromere:** It has a **submetacentric** centromere, meaning it has one arm slightly longer than the other.

Significant Genes Chromosome 5 houses several important genes involved in various biological functions, including: - **APC (Adenomatous Polyposis Coli):** A tumor suppressor gene associated with colorectal cancer. - **EGR1 (Early Growth Response 1):** A gene that plays a role in cell growth and differentiation. - **SLC9A3 (Sodium/Hydrogen Exchanger 3):** Important for kidney function and acid-base balance. - **SMN1 & SMN2 (Survival Motor Neuron 1 and 2):** Mutations in **SMN1** cause **Spinal Muscular Atrophy (SMA)**. - **TCOF1 (Treacle Protein 1):** Mutations in this gene lead to **Treacher Collins Syndrome**, a craniofacial disorder.

Associated Genetic Disorders Several genetic conditions and diseases are linked to mutations or deletions involving chromosome 5: - **Cri-du-chat Syndrome:** A disorder caused by a deletion of a portion of the short arm (5p), leading to intellectual disability, delayed development, and a characteristic "cat-like" cry in infants. - **Spinal Muscular Atrophy (SMA):** Caused by mutations in the **SMN1** gene, leading to progressive muscle weakness. - **Familial Adenomatous Polyposis (FAP):** A hereditary condition caused by mutations in the **APC** gene, leading to numerous precancerous polyps in the colon and a high risk of colorectal cancer.

Chromosomal Abnormalities - **5p Deletion Syndrome (Cri-du-chat Syndrome):** Caused by the loss of a portion of the short arm of chromosome 5. - **5q Deletion Syndrome (5q-Syndrome):** A condition associated with myelodysplastic syndromes (MDS), often leading to bone marrow failure.

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