Introduction

The human genome comprises 46 chromosomes arranged in 23 homologous pairs, of which 22 pairs are autosomes (Chromosomes 1–22) and one pair consists of the sex chromosomes. Each autosome encodes a distinct set of genes that govern critical biological processes ranging from embryonic development and immune function to neural architecture and metabolic regulation.

Numerical chromosomal abnormalities — either gain (trisomy, polysomy) or loss (monosomy, deletion) — disrupt gene dosage equilibrium, resulting in a spectrum of clinical presentations. This datasheet provides a structured reference of each autosome's primary biological role, associated pathological conditions, and the systemic effects of chromosomal imbalance.

Autosome Reference Compendium (Chromosomes 1 – 22)

1

CHROMOSOME

Largest human chromosome; regulates vital cellular functions including DNA repair, cell cycle control, and organ development.

ASSOCIATED CONDITIONS

Congenital Microcephaly, Gaucher Disease

2

CHROMOSOME

Second largest chromosome; plays a central role in neurological development, cognitive processing, and metabolic homeostasis.

ASSOCIATED CONDITIONS

Alström Syndrome

3

CHROMOSOME

Encodes the VHL tumor-suppressor gene; governs angiogenesis, hypoxia response, and renal cell biology.

ASSOCIATED CONDITIONS

Von Hippel-Lindau Disease

4

CHROMOSOME

Contains genes regulating bone and skeletal muscle formation, neural tube development, and craniofacial structure.

ASSOCIATED CONDITIONS

Huntington's Disease, Achondroplasia

5

CHROMOSOME

Controls cell proliferation, immune modulation, and development of multiple organ systems.

ASSOCIATED CONDITIONS

Cri-du-chat Syndrome

6

CHROMOSOME

Encodes the MHC/HLA immune-compatibility complex; central to adaptive immunity and transplant tolerance.

ASSOCIATED CONDITIONS

Autoimmune Disorders, Hemochromatosis

7

CHROMOSOME

Harbours the CFTR gene; contributes to language acquisition, neural circuit formation, and exocrine gland function.

ASSOCIATED CONDITIONS

Cystic Fibrosis, Williams Syndrome

8

CHROMOSOME

Regulates cell-cycle progression and contains the MYC proto-oncogene, critical for cell growth and proliferation.

ASSOCIATED CONDITIONS

Burkitt Lymphoma

9

CHROMOSOME

Carries the ABO blood-group locus and genes essential for DNA repair; supports biological maintenance pathways.

ASSOCIATED CONDITIONS

Galactosemia, Chronic Myeloid Leukemia (CML)

10

CHROMOSOME

Encodes the RET proto-oncogene; governs neural and endocrine gland development and neuroendocrine signalling.

ASSOCIATED CONDITIONS

Multiple Endocrine Neoplasia Type 2 (MEN2), Cowden Syndrome

11

CHROMOSOME

Contains key haematopoietic genes including the beta-globin cluster; regulates red blood cell structure and function.

ASSOCIATED CONDITIONS

Sickle Cell Anemia, Beta-Thalassemia

12

CHROMOSOME

Involved in enzyme biosynthesis, neural development, and motor neuron function throughout the central nervous system.

ASSOCIATED CONDITIONS

Phenylketonuria (PKU)

13

CHROMOSOME

Controls tissue growth, bone regeneration, and contains the RB1 retinoblastoma tumour-suppressor gene.

ASSOCIATED CONDITIONS

Patau Syndrome (Trisomy 13), Retinoblastoma

14

CHROMOSOME

Critical for immunoglobulin synthesis and adaptive immunity; harbours PSEN1, associated with early-onset Alzheimer's disease.

ASSOCIATED CONDITIONS

Autoimmune Conditions, Early-Onset Alzheimer's Disease

15

CHROMOSOME

Encodes genes governing metabolism, appetite regulation, motor skill development, and musculoskeletal coordination.

ASSOCIATED CONDITIONS

Prader-Willi Syndrome, Angelman Syndrome

16

CHROMOSOME

Regulates haemoglobin production and kidney tubular epithelium; contains the PKD1 polycystic kidney disease locus.

ASSOCIATED CONDITIONS

Polycystic Kidney Disease (PKD1)

17

CHROMOSOME

Harbours TP53 (guardian of the genome) and BRCA1; essential for tumour suppression and DNA-damage checkpoint control.

ASSOCIATED CONDITIONS

Neurofibromatosis Type 1 (NF1), Li-Fraumeni Syndrome

18

CHROMOSOME

Contains genes directing neural tube closure, organ morphogenesis, and early embryonic patterning.

ASSOCIATED CONDITIONS

Edwards Syndrome (Trisomy 18)

19

CHROMOSOME

Encodes APOE and genes governing cholesterol transport, liver metabolism, and neuromuscular junction function.

ASSOCIATED CONDITIONS

Myotonic Dystrophy

20

CHROMOSOME

Regulates tissue homeostasis, neural signal transduction, and encodes PRNP linked to prion protein biology.

ASSOCIATED CONDITIONS

Creutzfeldt-Jakob Disease

21

CHROMOSOME

Smallest autosome; critical for brain architecture, synaptic connectivity, and early cortical development.

ASSOCIATED CONDITIONS

Down Syndrome (Trisomy 21)

22

CHROMOSOME

Governs thymic development, myelination of neural tissue, and contains the NF2 tumour-suppressor locus.

ASSOCIATED CONDITIONS

DiGeorge Syndrome (22q11.2 Deletion), Neurofibromatosis Type 2

Clinical Effects of Chromosomal Numerical Imbalance

Chromosomal numerical imbalance — whether resulting from gain or loss of a whole chromosome or chromosomal segment — produces characteristic patterns of clinical manifestation. The following compendium outlines the principal phenotypic consequences observed across both categories of aneuploidy.

CHROMOSOMAL GAIN

- \+ Delayed physical growth and developmental milestones
- \+ Learning difficulties and reduced cognitive capacities
- \+ Speech, language, and communication delays
- \+ Congenital cardiac defects
- \+ Structural abnormalities across multiple organ systems
- \+ Nervous system disorders and neurological impairment
- \+ Poor gross and fine motor coordination
- \+ Hearing loss or visual impairment
- \+ Reproductive dysfunction and infertility
- \+ Elevated risk of syndrome-specific genetic disorders

CHROMOSOMAL LOSS

- – Severe developmental delay across all functional domains
- – Multiple congenital malformations
- – Impaired function of critical vital organs
- – Brain and central nervous system developmental disorders
- – Intellectual disabilities and significant learning challenges
- – Poor physical growth and proportional short stature
- – Hormonal and endocrine system abnormalities
- – Reproductive and fertility complications
- – Substantially increased risk of spontaneous miscarriage
- – In severe cases, incompatibility with postnatal life

Comparative Impact Summary

Clinical Domain	Chromosomal Gain (Trisomy)	Chromosomal Loss (Monosomy)
Cognitive Function	Mild–Moderate Impairment	Moderate–Severe Impairment
Physical Development	Delayed Milestones	Severe Growth Retardation
Organ Systems	Structural Malformations	Functional Organ Failure
Nervous System	Neurodevelopmental Delay	Severe Neural Dysgenesis

Reproductive Health	Reduced Fertility	Infertility / Absence
Cardiac System	Congenital Defects	Severe Cardiac Anomalies
Viability	Generally Viable	Often Lethal Prenatally