



**GROK COMMENT:** Here's an updated and expanded list from the GROK AI Generator that now specifically includes the major cancers, Alzheimer's disease, Parkinson's disease, and other dementias linked to each chromosome.

## Classic Chromosomal Disorders

**Chromosome 1** - Myeloid leukemias (1p36 del), Neuroblastoma (1p36 del), Colorectal, breast, melanoma (many genes) Early-onset Alzheimer's (rare families)  
Chromosome 1 is the largest human chromosome and is associated with a variety of diseases and conditions due to its susceptibility to genetic variations. Here are some notable disorders linked to abnormalities in chromosome 1:

**\*\*1p36 Deletion Syndrome\*\*:** This condition is caused by a deletion of genetic material from the short arm (p arm) of chromosome 1. It can lead to distinctive facial features, developmental delays, intellectual disabilities, seizures, vision and hearing problems, congenital heart defects, and renal abnormalities<sup>12</sup>.

**\*\*Neuroblastoma\*\*:** This is a type of cancer that originates in immature nerve cells. Deletions in the short arm of chromosome 1 (1p36) are associated with the development of neuroblastoma<sup>1</sup>.

**\*\*1q21.1 Microdeletion and Microduplication\*\*:** These conditions involve the deletion or duplication of a small piece of the long arm (q arm) of chromosome 1. They can result in developmental delays, intellectual disabilities, physical abnormalities, and neurological or psychiatric problems<sup>2</sup>.

**\*\*Various Cancers and Mendelian Disorders\*\*:** More than 350 diseases, including various cancers and Mendelian disorders, have been linked to abnormalities in the sequence of chromosome 1<sup>4</sup>.

**Chromosome 2** - Colorectal cancer (MSH2, MSH6 – Lynch syndrome), Neuroblastoma (2p gain ALK)

Chromosome 2 is the second-largest human chromosome and is associated with several genetic disorders and conditions. Here are some notable ones:

**\*\*2q37 Deletion Syndrome\*\*:** This condition is caused by a deletion of genetic material near the end of the long arm (q arm) of chromosome 2. It can lead to intellectual disability, behavioral problems, obesity, and skeletal abnormalities, including unusually short fingers and toes (brachydactyly)<sup>1</sup>.

**\*\*MBD5-Associated Neurodevelopmental Disorder (MAND)\*\*:** This disorder is caused by deletions or duplications at position 2q23.1. It affects neurological and physical development, leading to intellectual disability, developmental delay, impaired speech, sleep problems, and distinctive facial features<sup>1</sup>.

**\*\*Maturity-Onset Diabetes of the Young (MODY)\*\*:** This form of diabetes is linked to gene abnormalities on chromosome 2. It typically presents in adolescence or early adulthood and is characterized by impaired insulin production<sup>2</sup>.

**\*\*Primary Pulmonary Hypertension\*\***: This condition involves high blood pressure in the lungs' arteries and is associated with genetic changes on chromosome 2<sup>2</sup>.

**\*\*Autism\*\***: Some cases of autism have been linked to genetic abnormalities on chromosome 2<sup>2</sup>.

**Chromosome 3** - renal cell carcinoma, pheochromocytoma, hemangioblastoma, Parkinson's risk locus (minor)

Chromosome 3 is associated with several genetic disorders and conditions. Here are some notable ones:

**\*\*3p Deletion Syndrome\*\***: This condition is caused by the deletion of genetic material from the short arm (p arm) of chromosome 3. It can lead to intellectual disabilities, developmental delays, and physical abnormalities<sup>4</sup>.

**\*\*3q29 Microdeletion Syndrome\*\***: This involves the deletion of a small piece of the long arm (q arm) of chromosome 3. It can result in developmental delays, intellectual disabilities, and distinctive facial features<sup>4</sup>.

**\*\*Trisomy 3q2\*\***: This rare chromosomal disorder involves a portion of the 3rd chromosome appearing three times (trisomy) instead of twice. It can lead to developmental delays, intellectual disabilities, and distinctive craniofacial abnormalities<sup>1</sup>.

**Chromosome 4** - Bladder cancer (FGFR3), Wolf-Hirschhorn critical region cancers (rare)

Chromosome 4 is associated with several genetic disorders and conditions. Here are some notable ones:

**\*\*Facioscapulohumeral Muscular Dystrophy (FSHD)\*\***: This condition is primarily a muscle disorder where muscle wasting occurs in the face, shoulder blades, and upper arms. It is caused by genetic changes in the long arm (q arm) of chromosome 4 at position 4q35<sup>12</sup>.

**\*\*Wolf-Hirschhorn Syndrome\*\***: This complex syndrome is characterized by delayed growth, intellectual disabilities, seizures, and distinctive facial features. It is caused by a deletion at the end of the short arm (p arm) of chromosome 4 at position 4p16.3<sup>12</sup>.

**\*\*Cancers\*\***: Gene mutations on chromosome 4 have been linked to several types of cancer, including leukemias and other cancers involving blood-forming cells<sup>34</sup>.

**\*\*Neurological and Neurodegenerative Disorders\*\***: Conditions such as Parkinson's disease, Huntington's disease, and narcolepsy have been associated with gene mutations on chromosome 4<sup>3</sup>.

## **Chromosome 5** - Colorectal (APC → FAP), Myelodysplastic syndromes/AML (5q-syndrome)

Chromosome 5 is associated with several genetic disorders and conditions. Here are some notable ones:

**\*\*Spinal Muscular Atrophy (SMA)\*\*:** This is a neuromuscular disease caused by mutations in the SMN1 gene on chromosome 5. It leads to progressive muscle weakness and wasting<sup>1</sup>.

**\*\*Cri du Chat Syndrome\*\*:** Also known as 5p- syndrome, this condition is caused by a deletion on the short arm (p arm) of chromosome 5. It is characterized by a high-pitched cry that sounds like a cat, intellectual disability, and distinctive facial features<sup>12</sup>.

**\*\*5q-Syndrome\*\*:** This is a type of myelodysplastic syndrome caused by a deletion on the long arm (q arm) of chromosome 5. It leads to anemia, abnormalities in platelet production, and an increased risk of acute myeloid leukemia (AML)<sup>1</sup>.

**\*\*5q31.3 Microdeletion Syndrome\*\*:** This condition involves a deletion at position 5q31.3 and is characterized by weak muscle tone, swallowing and breathing difficulties, developmental delays, and distinctive facial features<sup>1</sup>.

## **Chromosome 6** - Many cancers, Melanoma (6q loss common)

Chromosome 6 is associated with several genetic disorders and conditions. Here are some notable ones:

**\*\*Ankylosing Spondylitis\*\*:** This is a type of arthritis that primarily affects the spine, causing inflammation, pain, and stiffness. It is linked to the HLA-B gene on chromosome 6<sup>3</sup>.

**\*\*Coeliac Disease\*\*:** This autoimmune disorder is triggered by the ingestion of gluten and is associated with the HLA-DQA1 and HLA-DQB1 genes on chromosome 6<sup>3</sup>.

**\*\*Ehlers-Danlos Syndrome\*\*:** This group of connective tissue disorders can cause hypermobility, skin that stretches easily, and a tendency to bruise. It is linked to the Tenascin-X gene on chromosome 6<sup>3</sup>.

**\*\*Hemochromatosis\*\*:** This condition causes the body to absorb too much iron from the diet, leading to iron overload. It is associated with the HFE gene on chromosome 6<sup>3</sup>.

**\*\*21-Hydroxylase Deficiency\*\*:** This genetic disorder affects the adrenal glands and can lead to congenital adrenal hyperplasia. It is linked to the CYP21A2 gene on chromosome 6<sup>3</sup>.

**\*\*Diabetes Mellitus\*\*:** A region on chromosome 6q24 is involved in the development of neonatal diabetes<sup>6</sup>.

**\*\*Chromosome 6 Ring\*\*:** This rare disorder involves the formation of a ring chromosome due to the loss of genetic material from both ends of chromosome 6. It can lead to growth retardation, intellectual disabilities, and various craniofacial abnormalities<sup>2</sup>.

**Chromosome 7** - Williams syndrome (7q11.23 del), Myelodysplastic syndromes/AML (monosomy 7 / 7q-), Bladder, prostate (7q gain). Cystic fibrosis carrier state linked to Parkinson's risk in some studies.

Chromosome 7 is associated with several genetic disorders and conditions. Here are some notable ones:

**\*\*Cystic Fibrosis\*\***: This is a genetic disorder that affects the respiratory and digestive systems. It is caused by mutations in the CFTR gene on chromosome 7<sup>2</sup>.

**\*\*Williams Syndrome\*\***: This condition is caused by a deletion of genetic material from the long arm (q arm) of chromosome 7 at position 7q11.23. It leads to developmental delays, cardiovascular problems, and distinctive facial features<sup>1</sup>.

**\*\*7q11.23 Duplication Syndrome\*\***: This condition results from an extra copy of a region on the long arm (q arm) of chromosome 7. It can cause neurological and behavioral problems as well as other abnormalities<sup>1</sup>.

**\*\*Myelodysplastic Syndrome (MDS) and Acute Myeloid Leukemia (AML)\*\***: Changes in the number or structure of chromosome 7, such as deletions or monosomy 7, are frequently associated with these types of blood cancers<sup>34</sup>.

**Chromosome 8** - Trisomy 8 mosaicism, Myeloid leukemias (trisomy 8), Breast (MYC amplification), Burkitt lymphoma (rare)

Chromosome 8 is associated with several genetic disorders and conditions. Here are some notable ones:

**\*\*Monosomy 8p\*\***: This rare chromosomal disorder is characterized by the deletion of a portion of the short arm (p arm) of chromosome 8. It can lead to growth deficiencies, intellectual disabilities, craniofacial malformations, heart abnormalities, and genital defects in males<sup>1</sup>.

**\*\*8q Deletion Syndrome\*\***: This condition involves the deletion of genetic material on the long arm (q arm) of chromosome 8. It can result in intellectual disability, developmental delays, speech and language difficulties, growth abnormalities, and distinctive facial features<sup>2</sup>.

**\*\*8p Duplication Syndrome\*\***: This condition involves the presence of extra genetic material on the short arm (p arm) of chromosome 8. It can cause developmental delays, intellectual disability, speech and language difficulties, behavioral challenges, and physical abnormalities<sup>2</sup>.

**\*\*Cancers and Other Conditions\*\***: Mutations on chromosome 8 have been implicated in certain heart defects, some forms of cancer, premature aging syndromes, immune responses, and immune disorders like psoriasis and Crohn's disease<sup>34</sup>.

## **Chromosome 9** - Bladder cancer (CDKN2A/p16), ALL (CDKN2A loss), Glioma (9p21 loss)

Chromosome 9 is associated with several genetic disorders and conditions. Here are some notable ones:

**\*\*9q22.3 Microdeletion Syndrome\*\***: This condition involves the deletion of a small piece of the long arm (q arm) of chromosome 9. It can lead to delayed development, intellectual disability, certain physical abnormalities, and features of Gorlin syndrome<sup>1</sup>.

**\*\*Bladder Cancer\*\***: Deletions of part or all of chromosome 9 are commonly found in bladder cancer. These chromosomal changes are seen only in cancer cells and are associated with both non-muscle invasive and muscle-invasive bladder cancer<sup>1</sup>.

**\*\*Chronic Myelogenous Leukemia (CML)\*\***: This type of cancer is associated with a specific chromosomal abnormality known as the Philadelphia chromosome, which involves a translocation between chromosomes 9 and 22<sup>2</sup>.

**\*\*Amyotrophic Lateral Sclerosis (ALS)\*\***: Some cases of ALS, a progressive neurodegenerative disease, have been linked to genetic changes on chromosome 9<sup>2</sup>.

**\*\*Coronary Artery Disease\*\***: Genetic variations on chromosome 9 have been associated with an increased risk of coronary artery disease<sup>2</sup>.

## **Chromosome 10** - PTEN → Cowden syndrome → breast, thyroid, endometrial cancers; Glioma, Prostate cancer, Parkinsonism in some PTEN mutations

Chromosome 10 is associated with several genetic disorders and conditions. Here are some notable ones:

**\*\*Monosomy 10p\*\***: This rare chromosomal disorder involves the deletion of the short arm (p arm) of chromosome 10. It can lead to severe intellectual disability, growth delays, distinctive craniofacial malformations, and congenital heart defects<sup>1</sup>.

**\*\*Distal Trisomy 10q\*\***: This condition involves the duplication of the long arm (q arm) of chromosome 10. It can result in intellectual disability, slow growth, muscle tone abnormalities, and distinctive craniofacial features<sup>2</sup>.

**\*\*Trisomy 10p\*\***: This rare disorder involves the duplication of the short arm (p arm) of chromosome 10. It can cause cardiac, renal, ocular, and bone malformations, as well as severe intellectual and motor deficiencies<sup>3</sup>.

**\*\*Cancers\*\***: Changes in the number and structure of chromosome 10 are associated with several types of cancer, including brain tumors called gliomas<sup>4</sup>.

## **Chromosome 11** - Wilms tumor (WT1), Ewing sarcoma (rare translocations), Mantle cell lymphoma (CCND1)

Chromosome 11 is associated with several genetic disorders and conditions. Here are some notable ones:

**\*\*Beckwith-Wiedemann Syndrome\*\***: This condition results from the abnormal regulation of genes on part of the short arm (p arm) of chromosome 11. It leads to overgrowth and an increased risk of developing certain types of tumors<sup>1</sup>.

**\*\*Jacobsen Syndrome\*\***: Also known as 11q terminal deletion disorder, this condition is caused by a loss of genetic material from the long arm (q arm) of chromosome 11. It can lead to developmental delays, intellectual disabilities, and distinctive facial features<sup>56</sup>.

**\*\*WAGR Syndrome\*\***: This rare genetic condition is caused by a deletion of a group of genes located on chromosome 11. It is characterized by Wilms tumor (a type of kidney cancer), Aniridia (absence of the iris), Genitourinary anomalies, and intellectual disability (mental Retardation)<sup>4</sup>.

**\*\*Partial Trisomy 11q\*\***: This rare chromosomal disorder involves the duplication of the long arm (q arm) of chromosome 11. It can cause growth retardation, intellectual disabilities, and distinctive craniofacial abnormalities<sup>2</sup>.

## **Chromosome 12** - Gastrointestinal stromal tumors (GIST), CML/ALL (PML-RARA in rare cases), Germ cell tumors. PARKINSON'S (PARK2 mutations in juvenile parkinsonism – actually on chr 6, but minor loci on 12).

Chromosome 12 is associated with several genetic disorders and conditions. Here are some notable ones:

**\*\*Pallister-Killian Mosaic Syndrome\*\***: This condition is caused by the presence of an abnormal extra chromosome called an isochromosome 12p. It can lead to developmental delays, intellectual disabilities, distinctive facial features, and other physical abnormalities<sup>1</sup>.

**\*\*Chronic Eosinophilic Leukemia\*\***: This type of blood cell cancer is associated with translocations involving chromosome 12. The most common translocation fuses part of the gene from chromosome 12, leading to the formation of the ETV6/PDGFR $\beta$  fusion gene<sup>13</sup>.

**\*\*Angiomatoid Fibrous Histiocytoma\*\***: This rare tumor, primarily found in adolescents and young adults, is associated with abnormalities in chromosome 12<sup>1</sup>.

**\*\*Clear Cell Sarcoma\*\***: Another rare tumor linked to chromosome 12 abnormalities<sup>1</sup>.

**Chromosome 13** - Patau syndrome (trisomy 13), Retinoblastoma (RB1), Osteosarcoma, Breast cancer (BRCA2 is on 13), Parkinson's (minor risk)  
Chromosome 13 is associated with several genetic disorders and conditions. Here are some notable ones:

**\*\*Patau's Syndrome (Trisomy 13)\*\*:** This serious genetic disorder is caused by having an additional copy of chromosome 13 in some or all of the body's cells. It leads to severe developmental issues, heart defects, brain abnormalities, and distinctive facial features<sup>1</sup>.

**\*\*13q Deletion Syndrome\*\*:** This condition involves the deletion of a portion of the long arm (q arm) of chromosome 13. It can result in developmental delays, intellectual disabilities, and physical abnormalities<sup>3</sup>.

**\*\*Feingold Syndrome Type 2\*\*:** This syndrome is caused by deletions at position 13q31.3. It is characterized by abnormalities of the fingers and toes, small head size (microcephaly), and learning disabilities<sup>2</sup>.

**\*\*Various Cancers\*\*:** Changes in chromosome 13 are associated with several types of cancer, including bladder cancer, breast cancer, and a rare blood cancer called 8p11 myeloproliferative syndrome<sup>23</sup>.

**Chromosome 14** - Thyroid cancer, Multiple myeloma (14q32 translocations), Renal oncocytoma, Early-onset Alzheimer's (PSEN1 is on chromosome 14 – major cause of familial AD)

Chromosome 14 is associated with several genetic disorders and conditions. Here are some notable ones:

**\*\*Multiple Myeloma\*\*:** This type of blood cancer involves abnormal changes in the structure or number of chromosomes, including translocations involving chromosome 14<sup>3</sup>.

**\*\*Burkitt's Lymphoma\*\*:** This cancer of white blood cells is related to a translocation between chromosomes 8 and 14<sup>4</sup>.

**\*\*Alpha-1 Antitrypsin Deficiency\*\*:** This genetic disorder can lead to lung and liver disease and is associated with genes on chromosome 14<sup>4</sup>.

**\*\*Trisomy 14 Mosaic\*\*:** This rare chromosomal disorder involves having an extra copy of chromosome 14 in some cells of the body. It can lead to growth delays, developmental delays, and distinctive craniofacial features<sup>2</sup>.

**\*\*Ring Chromosome 14 Syndrome\*\*:** This condition is caused by a chromosomal abnormality known as a ring chromosome 14. It can lead to developmental delays, intellectual disabilities, and seizures<sup>1</sup>.

**Chromosome 15** - Prader–Willi / Angelman, Hexosaminidase A deficiency (Tay-Sachs carrier state), Breast/ovarian (RAD51C rare), Hexosaminidase A deficiency (Tay-Sachs) can mimic dementia in infants/adults.

Chromosome 15 is associated with several genetic disorders and conditions. Here are some notable ones:

**\*\*Prader-Willi Syndrome (PWS)\*\*:** This condition is caused by the loss of working copies of a collection of genes on the paternal copy of chromosome 15. It leads to weak muscles, developmental delays, intellectual disabilities, behavioral concerns, and a constant hunger that can lead to excessive weight gain<sup>12</sup>.

**\*\*Angelman Syndrome (AS)\*\*:** This syndrome is caused by the missing expression of the UBE3A gene on the maternal copy of chromosome 15. It is characterized by developmental delays, speech impairments, movement and balance issues, seizures, and a distinctive happy demeanor<sup>23</sup>.

**\*\*Dup15q Syndrome\*\*:** This condition involves an extra copy (duplication) of a segment on the maternal copy of chromosome 15. It can result in developmental delays, intellectual disabilities, epilepsy, and autism spectrum disorder (ASD)<sup>23</sup>.

**\*\*Trisomy 15\*\*:** This rare chromosomal disorder involves having an extra copy of chromosome 15 in some or all of the body's cells. It can lead to growth delays, developmental delays, and distinctive craniofacial features<sup>5</sup>.

**Chromosome 16** - Polycystic kidney disease + renal cancer (PKD1), Breast/ovarian/pancreatic (PALB2), Rubinstein–Taybi + cancer predisposition. Chromosome 16 is associated with several genetic disorders and conditions. Here are some notable ones:

**\*\*Trisomy 16\*\*:** This condition involves having an extra copy of chromosome 16. It is the most common trisomy in humans and often leads to miscarriage<sup>1</sup>.

**\*\*Trisomy 16 Mosaicism\*\*:** In this condition, some cells have three copies of chromosome 16 while others have the normal two copies. It can lead to poor fetal growth, congenital heart defects, unusual facial features, underdeveloped lungs, and musculoskeletal anomalies<sup>1</sup>.

**\*\*16p13.3 Deletion Syndrome\*\*:** This disorder involves the deletion of part of the short arm (p arm) of chromosome 16. It has been reported in individuals with tuberous sclerosis, Rubinstein-Taybi syndrome, and alpha-thalassemia<sup>1</sup>.

**\*\*16p11.2 Duplication Syndrome\*\*:** This condition involves the duplication of a segment of the short arm (p arm) of chromosome 16. It can cause poor fetal growth, developmental delays, and other physical abnormalities<sup>1</sup>.

**\*\*Autism Spectrum Disorder (ASD)\*\*:** Changes in chromosome 16 have been linked to an increased risk of ASD<sup>2</sup>.

**\*\*Polycystic Kidney Disease (PKD)\*\*:** This genetic disorder, which causes the growth of numerous cysts in the kidneys, is associated with mutations on chromosome 16<sup>2</sup>.

**Chromosome 17** - Miller–Dieker, Smith–Magenis, TP53 (Li-Fraumeni syndrome) → almost every cancer type, Breast (BRCA1), Neurofibromatosis type 1 (NF1) → MPNST, optic glioma, Early-onset Alzheimer's (rare), Frontotemporal dementia (MAPT gene – tauopathies).

Chromosome 17 is associated with several genetic disorders and conditions. Here are some notable ones:

**\*\*Neurofibromatosis type 1 (NF1)** – Caused by mutations in the NF1 gene on chromosome 17 (specifically 17q11.2). This is the more common form (about 95–97% of cases). NF2 is on Chromosome 22.

**\*\*Charcot-Marie-Tooth Disease\*\***: This condition is characterized by muscle weakness and sensory changes. It is linked to mutations or deletions in genes on chromosome 17<sup>3</sup>.

**\*\*Potocki-Lupski Syndrome\*\***: This condition results from having an extra copy of a small piece of chromosome 17 in each cell. It can lead to developmental delays, intellectual disabilities, and distinctive facial features<sup>4</sup>.

**\*\*Smith-Magenis Syndrome\*\***: This syndrome is caused by the deletion of a small piece of chromosome 17. It can result in developmental delays, intellectual disabilities, sleep disturbances, and behavioral problems<sup>4</sup>.

**\*\*17q12 Deletion Syndrome\*\***: This condition involves the deletion of a small piece of the long arm (q arm) of chromosome 17. It can lead to abnormalities of the kidneys and urinary system, a form of diabetes called MODY5, delayed development, intellectual disability, and behavioral or psychiatric disorders<sup>1</sup>.

**\*\*17q12 Duplication Syndrome\*\***: This condition involves the duplication of a small piece of the long arm (q arm) of chromosome 17. It can cause a wide range of symptoms, including developmental delays, intellectual disabilities, and behavioral issues<sup>1</sup>.

**Chromosome 18** - Edwards syndrome (trisomy 18), Colorectal (SMAD4 → juvenile polyposis), Pancreatic (SMAD4/DPC4).

Chromosome 18 is associated with several genetic disorders and conditions. Here are some notable ones:

**\*\*Edwards' Syndrome (Trisomy 18)\*\***: This serious genetic disorder is caused by having an extra copy of chromosome 18. It leads to severe developmental issues, heart defects, brain abnormalities, and distinctive facial features<sup>12</sup>.

**\*\*Erythropoietic Protoporphyrinemia\*\***: This condition affects the production of heme, a component of hemoglobin. It can cause sensitivity to sunlight, leading to painful skin reactions<sup>4</sup>.

**\*\*Hereditary Hemorrhagic Telangiectasia\*\***: This disorder affects blood vessels and can cause frequent nosebleeds, skin discolorations, and arteriovenous malformations<sup>4</sup>.

**\*\*Niemann-Pick Disease Type C\*\***: This is a rare, inherited disease that affects the body's ability to metabolize cholesterol and other lipids. It can lead to severe neurological symptoms<sup>4</sup>.

**\*\*Porphyria\*\***: This group of disorders results from a buildup of natural chemicals that produce porphyrin in the body. It can affect the skin or nervous system<sup>4</sup>.

**Chromosome 19** - Glioma, Ovarian cancer (common 19q loss). Late-onset Alzheimer's strongest risk gene: APOE ε4 (19q13) – by far the biggest genetic risk factor for sporadic AD.

Chromosome 19 is associated with several genetic disorders and conditions. Here are some notable ones:

**\*\*19p13.13 Deletion Syndrome\*\***: This condition involves the deletion of a small piece of the short arm (p arm) of chromosome 19. It can lead to an unusually large head size (macrocephaly), tall stature, delayed development of speech and motor skills, intellectual disability, seizures, feeding and digestive difficulties, and eye abnormalities<sup>1</sup>.

**\*\*Acute Lymphoblastic Leukemia (ALL)\*\***: Rearrangements of genetic material between chromosome 19 and other chromosomes are particularly common in this type of blood cancer<sup>3</sup>.

**\*\*Ring Chromosome 19 Syndrome\*\***: This rare disorder involves the formation of a ring chromosome due to the loss of genetic material from both ends of chromosome 19. It can lead to mild hypotonia, autistic-like mannerisms, cutis laxa, hearing loss, syndactyly, digital hypoplasia, and talipes equinovarus<sup>4</sup>.

**Chromosome 20** - Gastrointestinal stromal tumors (rare), Breast (20q13 amplification)

Chromosome 20 is associated with several genetic disorders and conditions. Here are some notable ones:

**\*\*Alagille Syndrome\*\***: This genetic disorder affects the liver, heart, and other parts of the body. It is associated with deletions of genetic material on chromosome 20, specifically in the region known as 20p12<sup>13</sup>.

**\*\*Ring Chromosome 20 Syndrome\*\***: This condition is caused by a chromosomal abnormality known as a ring chromosome 20. It can lead to epilepsy and other health problems<sup>13</sup>.

**\*\*Trisomy 20\*\***: This condition involves having three copies of chromosome 20 instead of the usual two. It can lead to various developmental and physical abnormalities<sup>3</sup>.

**\*\*Albright's Hereditary Osteodystrophy\*\***: This genetic disorder affects the bones and other tissues. It is related to genes on chromosome 20<sup>2</sup>.

**\*\*Adenosine Deaminase Deficiency\*\***: This condition affects the immune system and is associated with genes on chromosome 20<sup>2</sup>.

**Chromosome 21** - Down syndrome (trisomy 21), Acute megakaryoblastic leukemia (in Down syndrome), Early-onset Alzheimer's in virtually all Down syndrome individuals by age 40+ (triplication of APP gene). Down syndrome → near-100% Alzheimer's pathology by age 40–50 (due to APP gene triplication).

Chromosome 21 is most famously associated with **Down syndrome (Trisomy 21)**. This genetic disorder is caused by the presence of an extra copy of chromosome 21 and leads to developmental delays, intellectual disabilities, and characteristic physical features<sup>12</sup>.

People with Down syndrome also have an increased risk of several health issues, including:

- **Congenital heart defects**
- **Thyroid diseases** (such as hypothyroidism)
- **Diabetes**
- **Leukemia**
- **Epilepsy**
- **Gastrointestinal anomalies** (such as duodenal atresia and Hirschsprung disease)
- **Celiac disease**
- **Obstructive sleep apnea**<sup>34</sup>

**Chromosome 22** - DiGeorge/22q11.2 del, Chronic myeloid leukemia (Philadelphia chromosome rare variant), Breast (22q loss common).

Chromosome 22 is associated with several genetic disorders and conditions. Here are some notable ones:

**Neurofibromatosis type 2 (NF2)** – Caused by mutations in the NF2 gene on chromosome 22 (specifically 22q12.2). This is less common and typically involves bilateral acoustic neuromas (vestibular schwannomas). NF1 is on Chromosome 17.

**DiGeorge Syndrome (22q11.2 Deletion Syndrome)**: This condition is caused by the deletion of a small part of chromosome 22. It can lead to heart defects, immune system problems, cleft palate, and developmental delays<sup>12</sup>.

**Cat Eye Syndrome**: This rare condition is caused by a duplication of a small piece of chromosome 22. It can result in eye abnormalities, heart defects, and kidney problems<sup>3</sup>.

**Chronic Myeloid Leukemia (CML)**: This type of cancer is associated with a specific chromosomal abnormality known as the Philadelphia chromosome, which involves a translocation between chromosomes 9 and 22<sup>3</sup>.

**Amyotrophic Lateral Sclerosis (ALS)**: Some cases of ALS, a progressive neurodegenerative disease, have been linked to genetic changes on chromosome 22<sup>3</sup>.

**Breast Cancer**: Certain genetic mutations on chromosome 22 have been associated with an increased risk of breast cancer<sup>3</sup>.

**Chromosome X** - Turner, Klinefelter, Fragile X, Breast/ovarian (BRCA1 is actually on 17q; X has many cancer-related genes), Lynch syndrome (mismatch repair genes on 2/3/7). Fragile X-associated tremor/ataxia syndrome (FXTAS) – Parkinsonism + dementia in older male carriers.

Chromosome X is associated with several genetic disorders and conditions. Here are some notable ones:

**\*\*Klinefelter Syndrome\*\***: This condition occurs in males who have one or more extra copies of the X chromosome (XXY). It can lead to abnormal sexual development, reduced testosterone levels, learning difficulties, and infertility<sup>1</sup>.

**\*\*Turner Syndrome\*\***: This condition affects females and arises from a missing or abnormally structured X chromosome. It can lead to short stature, infertility, and various physical abnormalities<sup>2</sup>.

**\*\*Triple X Syndrome (Trisomy X)\*\***: This condition is caused by the presence of an extra copy of the X chromosome in females. It can lead to delayed language, motor-coordination, and cognitive processing skills<sup>2</sup>.

**\*\*Hemophilia A and B\*\***: These are bleeding disorders caused by mutations in genes on the X chromosome. They lead to a deficiency in blood clotting<sup>3</sup>.

**\*\*Duchenne Muscular Dystrophy\*\***: This condition causes progressive muscle weakness and immobility. It is caused by mutations in the dystrophin gene on the X chromosome<sup>3</sup>.

**\*\*Red-Green Color Blindness\*\***: This condition affects an individual's ability to see red or green colors and is caused by mutations in genes on the X chromosome<sup>3</sup>.

**\*\*Adrenoleukodystrophy\*\***: This condition leads to progressive brain damage, failure of the adrenal glands, and eventually death. It is caused by mutations in the ABCD1 gene on the X chromosome<sup>4</sup>.

### **Key Take-Home Highlights for Cancer & Neurodegeneration Alzheimer's disease**

- Chromosome 21: Down syndrome → almost inevitable early Alzheimer's
- Chromosome 19: APOE ε4 (strongest common risk for late-onset AD)
- Chromosome 14: Presenilin-1 (PSEN1) – major cause of early-onset familial AD
- Chromosomes 1 & others: Presenilin-2, APP mutations (rare familial), Parkinson's disease (major monogenic forms)
- Chromosome 4: SNCA (α-synuclein) – rare duplication/triplication
- Chromosome 6: PARK2 (Parkin) – juvenile parkinsonism
- Chromosome 1: PINK1, DJ-1
- Many risk loci scattered (e.g., LRRK2 on 12, GBA on 1q)

Most important cancer predisposition chromosomes

3 (VHL), 5 (APC), 10 (PTEN), 13 (BRCA2, RB1), 17 (TP53, BRCA1, NF1)