List of Diseases Caused by Chromosomal Disorder

Today we are presenting you a short note on diseases caused by chromosomal disorder which is very important for RRB NTPC 2016, SSC CGL 2016 and other upcoming exams.

Diseases Caused by Chromosomal Disorder

Normally, humans have 23 pairs of chromosomes – making 46 in total. This includes one pair of chromosomes which are the sex chromosomes. The ova and the sperm each carry 23 chromosomes.

Chromosomal abnormalities occur when there is a defect in a chromosome, or in the arrangement of the genetic material on the chromosome. Very often, chromosome abnormalities give rise to specific physical symptoms, however, the severity of these can vary from individual to individual. Here the list of the diseases with chromosomal disorder and symptoms.

Name of the Disease	Chromosome Disorder	Symptoms/ Disorder
22q11.2 Deletion Syndrome	deletion of a small piece of chromosome 22 near the middle of the chromosome	DiGeorge syndrome, velocardiofacial syndrome (also called Shprintzen syndrome), and conotruncal anomaly face syndrome
Angelman Syndrome	deletion or inactivitation of genes on the maternally inherited chromosome 15	intellectual and developmental delays, sleep disturbances, seizures, and jerky movements, but also frequent laughter or smiling and usually have a happy demeanor
Cat Eye Syndrome	the short arm (known as 22p) and a small region of the long arm (22q) of chromosome 22 are present three or four times, rather than twice	mild growth delays before birth, mild mental deficiency, and malformations of the skill and facial region, the heart, the kidneys, and/or the anal region
Charcot-Marie-Tooth Disease	duplication of the gene on chromosome 17	Weakness in your legs, ankles and feet. Loss of muscle bulk in legs and feet. High foot arches. Curled toes (hammertoes). Decreased ability to run. Difficulty lifting your foot at the ankle (footdrop). Awkward or higher than normal step (gait)

Cri du Chat Syndrome	Missing a piece of chromosome 5.	a high-pitched cry that sounds like a cat, downward slant of the eyes, partial webbing or fusing of fingers or toes, and slow or incomplete development of motor skills
Jacobsen Syndrome	loss of genetic material from the end of the long arm of chromosome 11	signs and symptoms of this condition vary, but most individuals experience delayed development in motor skills and speech, cognitive impairments, learning difficulties, and some behavioral problems.
Trisomy 13/Patau Syndrome	three copies of genetic material from chromosome 13	heart defects, brain or spinal cord abnormalities, very small or poorly developed eyes (microphthalmia), extra fingers or toes, an opening in the lip (a cleft lip) with or without an opening in the roof of the mouth (a cleft palate), and weak muscle tone (hypotonia)
Trisomy 16	three copies of chromosome 16 instead of the usual two	intrauterine growth retardation (IUGR) and congenital heart defects
Trisomy 18/Edwards Syndrome	third copy of material from chromosome 18 instead of the usual two copies	clenched hands, feet with a rounded bottom, mental deficiency, underdeveloped fingernails, and an unusual shaped chest
Triple X Syndrome	an extra X chromosome in each of a female's cells	it does not cause any unusual physical features but is associated with the increased risk of learning disabilities and delayed development of speech and language skills
Turner Syndrome	one of the two X chromosomes in females is either missing or incomplete	short stature and gonadal dysgenesis, which can cause incomplete sexual development and ovarian failure and infertility
Williams Syndrome	a deletion of genetic material from portions of the long arm of chromosome 7	the symptoms of Williams Syndrome is still unknown

Wolf-Hirschhorn Syndrome	deletion of the distal short arm of chromosome 4	facial appearance, delayed growth and development, intellectual disability, and seizures