Human Genetics.

There are certain defects which are caused due to genetic defects disorders, which may be due to Mendelian genes, multifactor genes, chromosomal abberations and anomalies.

(1) Sickel-Cell Anaemia :- It is caused due to a defective allele (gene) on 11th chromosome (chromosome no. 11. → Autosome). It is recessive. In homozygous condition individual do not survive, but in heterozygous it leads to haemolytic anaemia under condition of deficiency of oxygen. If the other allele is normal it causes 60-70% of normal haemoglobin. This abnormality is due to replacement of glutamine at 6th place of β-chain (Globin of haemoglobin molecule) by Valine. Due to deficiency of oxygen R.B.C.s become sickel shaped due to elongation. They move slowly causing clumping in capillaries on venous side and then get distroyed. Later on it may cause (i) damage of tissues and even organs and (ii) Haemolytic anemia. It may cause ulceration on the lower extremities, attacks of abdominal pain, joint pains. It mainly causes deficient transport of oxygen, food, and waste products which leads to defects of various organs.

[2] Phenylketonuria (PKU) :- It is caused due to recessive defective allele on chromosome no. 12 (12th chromosome → Autosome). Due to this phenylalanine hydroxylase is not formed. It causes accumulation of phenylalanine and its derivatives like, phenyl acetate, phenyl pyruvate and phenyl lactate in cerebro blood, and cerebro-spinal fluid causing mental retardation. It causes, Eczema, hypo/hyper pigmentation, mousy odour (brown, white & hair).

~~This disease can be managed due to~~
The child born normal. The effects are seen after few weeks. It may be managed by giving diet which lacks phenylalanine.

[3] ~~AD~~ Alzheimer's Disease :- It is caused due to two abnormal / defective genes present separately on chromosome no. 19 and ~~the~~ no. 21.

The E4 gene of chromosome no. 19. causes the disease in late forties (after the age of 40).

The defect of chr-21 is dominant and causes the disease in early age. It is rare and associated with ~~down~~ Down's Syndrome.

In this disease degeneration of neurons takes place due to deposition of senile plaques caused by non-metabolism of amyloid β-peptide. Activity of choline acetyl transferase is reduced.

It leads in trembling of hand, change in personality, disturbance in memory, ~~which~~ increases progressively.
Loss of memory

DOWN'S SYNDROME :-

Caused due to trisomy that is one more chromosome (3 of chrom. 21) instead of 2 of ch 21.

It is non disjunction done during gametogenesis i.e. $(n+1) \times (n)$.

It takes place in late age pregnancy (35-40) or lower age of mother (14-16) below 18 and higher age of father (50 and above).

This disease is discovered by Langdon Down in 1866. and named after him. Its In 1959 J. Lejeune discovered its cause.

The patients have short stature, small round heads, broad forheads, small ears, open mouth, projecting lower lip, protruding furrowed tongue, epicanthal fold on each side of nose, short flat bridge nose, short neck, short broad hands, short phalanges, space between first and second digit, moderate to severe mental retardation, retarded physical and physiomotor development (loose jointedness, underdeveloped gonads and genitals) and short life expectancy.

In rare cases it is caused due to translocation of a part of 21 chr. no. to chr. no. 14. In such case the chr. no. remain 46. but partial trisomy due to additional part of ~~on chr. no.~~ 14. of chr. no. 21. of chr. no. 14. It is known as Familial Down's Syndrome.

[4] Patau's Syndrome: It occurs due to trisomic for chromosome no. 13. It occurs due to non-disjunction of this chromosome during gametogenesis. One of the gamete contain (n+1) where other is normal.

The affected children show - defective eyes, defective heart and mental retardation. Cleft palate & lip, polydactyly, anomalous heart viscera, genitalia, and dermal pattern.

[5] Edward's Syndrome:- It is caused due to an inborn defect i.e. the trisomy for chromosome no. 18. It is caused due to non-disjunction during gametogenesis. One of the gametes contain (n+1) where other is normal.

The affected child shows anomalies of fingers, complex digital prints, defects of heart, low set ears, small mouth, long narrow skull. Such a patient generally dies within one year of its birth.

Such a child has narrow skull, small face, corneal opacity, webbed neck etc.