Perspective

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Chromosomal Abnormalities such as Translocation of the Genetic Material

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Description

The development of the nervous system is tightly regulated and timed; it is influenced by both genetic programs and the environment. Any significant deviation from the normal developmental trajectory early in life can result in missing or abnormal neuronal architecture or connectivity. Because of the temporal and spatial complexity of the developmental trajectory, there are many potential causes of neurodevelopmental disorders that may affect different areas of the nervous system at different times and ages. These range from social deprivation, genetic and metabolic diseases, immune disorders, infectious diseases, nutritional factors, physical trauma, and toxic and environmental factors.

Some neurodevelopmental disorders, such as autism and other pervasive developmental disorders are considered multifactorial syndromes which have many causes that converge to a more specific neurodevelopmental manifestation. A prominent example of a genetically determined neurodevelopmental disorder is Trisomy 21, also known as Down syndrome. This disorder usually results from an extra chromosome 21, although in uncommon instances it is related to other chromosomal abnormalities such as translocation of the genetic material. It is characterized by short stature, epicanthal (eyelid) folds, abnormal fingerprints, and palm prints, heart defects, poor muscle tone delay of neurological development, and intellectual disabilities delay of intellectual development.

Variant Disorder

Less commonly known genetically determined neuro developmental disorders include Fragile X syndrome. Fragile X syndrome was first described in 1943 by Martin and Bell, studying persons with family history of sex-linked mental defects. Ret syndrome, another X-linked disorder, produces severe functional limitations. Williams's syndrome is caused by small deletions of genetic material from chromosome 7. The most common recurrent copy number variant disorder is deletion syndrome formerly diverge or velocardiofacial syndrome, followed by Prader-Willi syndrome and angel man syndrome. Immune reactions during pregnancy, both maternal and of the developing child, may produce neurodevelopmental disorders. One typical immune reaction in infants and children is "pandas" or Pediatrics Autoimmune Neuropsychiatric Disorders Associated with Streptococcal infection. Another disorder is Sydenham's chorea, which results in more abnormal movements of the body and fewer psychological sequellae. Both are immune reactions against brain tissue that follow infection by Streptococcus bacteria. Susceptibility to these immune diseases may be genetically determined, so sometimes several family members may have one or both of them following an epidemic of Strep infection. citation need Metabolic disorders in either the mother or the child can cause neurodevelopmental disorders. Two examples are diabetes mellitus a multifactorial disorder and phenylketonuria an inborn error of metabolism. Many such inherited diseases may directly affect the child's metabolism and neural development but less commonly they can indirectly affect the child during gestation. See also teratology. In a child, type 1 diabetes can produce neurodevelopmental damage by the effects of excess or insufficient glucose. The problems continue and may worsen throughout childhood if the diabetes is not well controlled. Type 2 diabetes may be preceded in its onset by impaired cognitive functioning. A non-diabetic foetus can also be subjected to glucose effects if its mother has undetected gestational diabetes. Maternal diabetes causes excessive birth size, making it harder for the infant to pass through the birth canal without injury or it can directly produce early neurodevelopmental deficits. Usually the neurodevelopmental symptoms will decrease in later childhood. Phenylketonuria, also known as PKU, can induce neurodevelopmental problems and children with PKU require a strict diet to prevent intellectual disability and other disorders. In the maternal form of PKU, excessive maternal phenylalanine can be absorbed by the foetus even if the foetus has not inherited the disease. This can produce intellectual disability and other disorders. Nutrition disorders and nutritional deficits may cause neurodevelopmental disorders, such as spin bifida, and the rarely occurring anencephaly, both of which are neural tube defects with malformation and dysfunction of the nervous system and its supporting structures, leading to serious physical disability and emotional squeal. The most common nutritional cause of neural tube defects is folic acid deficiency in the mother, a B vitamin usually found in fruits, vegetables, whole grains, and milk products.

Intellectual Disability

Neural tube defects are also caused by medications and other environmental causes, many of which interfere with floated metabolism, thus they are considered to have multifactorial causes. Another deficiency, iodine deficiency, produces a spectrum of neurodevelopmental disorders ranging from mild emotional disturbance to severe intellectual disability. See also congenital iodine deficiency syndrome. Excesses in both maternal and infant diets may cause disorders as well, with foods or food supplements proving toxic in large amounts. For instance in 1973 K.L. Jones and D.W. Smith of the University of Washington Medical School in Seattle found a pattern of "craniofacial, limb, and cardiovascular defects associated with prenatal onset growth deficiency and developmental delay" in children of alcoholic mothers, now called foetal alcohol syndrome, It has significant symptom overlap with several other entirely unrelated neurodevelopmental disorders Neurodevelopmental disorders are diagnosed by evaluating the presence of characteristic symptoms or behaviours in a child, typically after a parent, guardian, teacher, or other responsible adult has raised concerns to a doctor.

Neurodevelopmental disorders may also be confirmed by genetic testing. Traditionally, disease related genetic and genomic factors are detected by karyotype analysis, which detects clinically significant genetic abnormalities for 5% of children with a diagnosed disorder. As of 2017, Chromosomal Microarray Analysis (CMA) was proposed to replace karyotyping because of its ability to detect smaller chromosome abnormalities and copy-number variants, leading to greater diagnostic yield in about 20% of cases.