Genetics and fetal health

Courtesy of Prof. Shane Higgins' website

This article about genetics and fetal health serves as an introduction to the role of genetics in fetal medicine. It is intended to offer readers a basic understanding of the subject matter.

1. What is fetal medicine?

Fetal medicine is a relatively new super-specialised branch of medicine that manages the evaluation of fetal growth and timely identification of congenital disabilities. It is a sub-speciality of obstetrics that concentrates on the diagnosis and treatment of expectant mothers and their unborn infants. The maternal physicians specialise in providing care for females who are at high risk for problems during their pregnancies. [1] Part of this process can include understanding and diagnosing the role of genetics in fetal medicine.

Most parents hope to give birth to healthier and medically fit babies. Most babies born to women of all ages are healthy and normal. Regrettably, some pregnancies are at risk for congenital disabilities or genetic problems. (If you wish to understand more about the normal journey of fetal development, then this article about Fetal Development Stages Month by Month may be of interest to you.)

There is a small chance for these issues in any pregnancy, but also the risks might be more significant due to some conditions. Even before your pregnancy, the health care provider may rate your health risks by taking a questionnaire to evaluate prior health and pregnancy history. Before and throughout your pregnancy, your healthcare provider will suggest some screening tests to determine if you may be at increased risk for congenital disabilities or inherited circumstances that have the potential to affect your unborn baby.

2. What is the role of genetics in fetal medicine

Sometimes the genes may contain contradictory instructions, and generally, one gene will win the struggle for dominance. Some genes behave in an additive manner. For instance, if a young child has one tall parent and a short parent, their child would be of average height.

In other cases, some genes follow the dominant-recessive routine of gene expression. Parent's genes predetermine the limits of a person's height and other physical features like the variability in eye colour, hair colour, body make-up, and skin tone. When two parents are homogeneous to get a blonde hair allele, they cannot have a black-hair child, but if the two parents are heterozygous and have a black hair allele, then there is a 25% percent probability their child will inherit homozygous alleles that produce a phenotype for dark hair.

3. What are the types of genetic abnormalities?

There are several varieties of hereditary disorders. The way in which a disease is inherited will help determine the dangers it will have for a pregnancy and the possibility of recurrence; if it is going to replicate in future children.

Several factors depend upon the genetic abnormalities such as if the parents have a chromosomal abnormality. Secondly, the family history of genetic disorders. Thirdly, if parents have given birth to a genetically abnormal child. Lastly, if the unborn fetus carries genetic abnormalities which are confirmed by ultrasound.

The genetic directions guided by DNA is a never failing process. There are lots of varieties of hereditary disorders. The pattern by which the disease is inherited will influence the dangers it is going to have in pregnancy and the chance of recurrence it is going to replicate in the offspring. When the abnormal cells combine with a normal cell, the resulting zygote is going to have an uneven number of chromosomes. Researchers indicate as many as half of zygotes which form have less or more than 23 chromosomes, but the majority of them are spontaneously aborted rather than grow to a full-term infant. [2]

Types of abnormalities due to genes

Achondroplasia

Achondroplasia is a bone growth illness which leads to disproportionate dwarfism. Dwarfism is described as a state of short height as a grown-up. Individuals with achondroplasia reach full height, with a regular sized chest but the limbs that are short. It is the most typical kind of disproportionate dwarfism. It is an autosomal dominant hereditary trait; achondroplasia happens as a result of a new spontaneous change in genetic material in about 90 percent of cases. [3]

Marfan Syndrome

Marfan syndrome is a genetic disorder which affects the body's connective tissues. Connective tissue retains all of the body's cells, tissue and organs together. Also, it has a significant function in assisting your body in the developmental stages. Marfan syndrome is an inherited connective tissue disease noteworthy because of its global distribution and comparatively higher incidence. [4]

4. What are the types of chromosomal abnormalities?

Chromosomes are stick-shaped constructions at the centre of every cell within the entire body. If a chromosome is strange, it can lead to health problems within the body. Chromosome abnormalities most often occur as a consequence of an error during cellular division.

Types of Chromosomal Abnormalities

Aneuploidy

Aneuploidy is one of the significant sorts of chromosome mutations where the chromosome number is unnatural. An aneuploid is a person whose chromosome amount differs in the 46 by a part of a chromosome collection. Typically, the aneuploid chromosome collection is different from 46 by just one or a few chromosomes. Aneuploids may have a chromosome number either larger or smaller compared to 46. Aneuploid nomenclature is determined by the number of duplicates of the particular chromosome from the aneuploid state.[5]

Down Syndrome

Down Syndrome is the most typical numerical chromosomal abnormality. Also known as trisomy 21, Down Syndrome is a chromosomic disorder produced by the existence of part or another third copy of chromosome 21. This syndrome is one of the most common genetic abnormalities. Important physical and intellectual disabilities represent the patients with this syndrome.

Patients may have a multiple list of signs and symptoms such as: slanted eyes, poor muscle tone, single crease of the palm, macroglossia, flat and wide face, short neck, toe malformations, short stature, low IQ levels (adult patients with the mentality of a child), delayed developmental milestones, risk of epilepsy, hearing and vision disorders, congenital cardiac, endocrine, gastrointestinal and reproductive issues.

Diagnosis can be made by ultrasound, amniocentesis, chorionic villus sampling and symptomatology. There is no cure for Down Syndrome, but the patients can receive education and proper care, to attain a good quality of life.

Turner Syndrome

Also known as 45,X, Turner Syndrome is a pathology that affects only women and is based on the partial or complete absence of an X chromosome in some or all of the cells, leading to usually having 45 chromosomes. Almost 99% of fetuses with this syndrome spontaneously terminate during the first three months of pregnancy.

Patients may have a multiple list of signs and symptoms such as: broad chest, ears located lower than normal, short stature, swelling of the hands and feet, reproductive sterility, underdeveloped ovaries, amenorrhoea, absence of sexual maturation, obesity, webbed neck, cardiac malformations, horseshoe kidney, endocrine disorders, ophthalmic issues, attention deficit hyperactivity disorder, among other cognitive disorders. Diagnosis can be made by amniocentesis or chorionic villus sampling or abnormal imaging tests findings during pregnancy.

After birth, Turner Syndrome can be diagnosed based on symptomatology or karyotype. There is no cure for this pathology, but much can be done to minimise symptoms: growth hormone, oestrogen replacement therapy and reproductive technologies. [7]

Diagnostic Procedure

Families having a risk for hereditary diseases might be referred to a certified Genetic Counsellor. The family's medical background might help determine risks for specific issues. Genetic counselling helps parents understand that the ramifications of disease and ways it could be avoided or treated. It could be required to look at each parent's DNA to find out about a few genetic inheritance patterns. Prenatal testing is also available to inspect the fetus. It may consist of ultrasound, chorionic villus sampling, and amniocentesis.

For the most up to date information about genetic counselling and testing in Dublin, please refer to the UCD information about Genetic Counselling for Genetic Disorders.

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