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Pathological Analysis of down Syndrome and Its Nursing Care

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Abstract. Down Syndrome sounds like a rare case that can only be found in biology courses that are unrelated to normal people's life. However, it occurs once every 1200 people according to CDC. Scientifically, Down syndrome is a genetic disease caused by trisomy 21 that cannot be cured by direct means of surgery or drugs. People must study the biochemical pathway Down Syndrome interferes with human body development, symptoms usually exhibit in patients, and Down Syndrome's relationship with other diseases like Alzheimer's in-depth and provide advanced detection for parents inclined to have Down Syndrome babies. Due to patients' intellectual disability, someone must make important decisions for them so that they can receive needed treatments, Nursing patients with it not only requires extensive medical care but also patient emotional support. Well-trained nursing is required to protect this vulnerable group, not only to bring patients the chance to live a normal life but also to help their families avoid living in conflicts.

Keywords: down syndrome, nursing care, pathological analysis

1. Introduction

Down syndrome is a chromosome disorder caused by three copies of chromosome 21. Commonly, the human genome is composed of 23 pairs of chromosomes, 22 pairs of autosomes, and one set of sex chromosomes. Due to errors that occur during mitosis or meiosis, patients with Down syndrome have an extra 21 chromosome [1].

Down syndrome is different from infectious diseases which are caused by viruses or bacteria since what goes wrong is the chromosome-the carrier of genetic information. Down syndrome can only be inherited, but it also differs from other genetic diseases since it is not a special gene mutation passed by parents but an error that happens during cell division.

This paper mainly discusses the causes of Down syndrome, and talks about the ways currently used to deal with the Down syndrome, so as to offer some references for future studies in the treatment and nursing care of Down syndrome.

2. Pathological analysis of down syndrome

2.1. Causes of down syndrome

There are simply three ways of chromosomal changes that Down Syndrome can be caused.

First, if chromosome 21 does not separate during meiosis, some of the gametes produced will have one extra chromosome. By combining with the chromosomes that already exist, the gamete owns two

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copies of the same chromosome. When two gametes fuse, this is something harmful to the formation of sperm or ova: it ends up with a germ cell that has three copies of the same chromosome. This path of cause is scientifically named complete Trisomy 21 [2].

Second, a rare form of Down Syndrome named Mosaic trisomy 21 which occurs in less than five percent of the cases is caused by abnormal cell division after fertilization. “Mosaic” here simply means “combination”, patients with Mosaic trisomy have some of their somatic cells healthy while others have three copies of chromosome 21. It is a combination of normal cells and abnormal ones. Different from Trisomy 21, since the patients have normal somatic cells, the error goes wrong after two gametes form: sick cells can only produce offspring that display trisomy. The degree to which patients exhibit symptoms depends on the percentage of trisomy cells [3].

Patients with the last type of three chromosomal changes, Translocation trisomy 21, show similar symptoms to those of complete trisomy 21. They have their extra chromosome 21 attached to other chromosomes rather than the normal pair of 21s. Trisomy is an exhibit not on chromosome 21 but on others [4].

2.2. The biochemistry reasons for the symptoms

So how can trisomy damage one’s brain? There are two famous perspectives regarding this question: the gene dosage effect and the amplified developmental instability. They all focused on the way overexpression of special genes in chromosome 21 affect our neuron.

Gene dosage effects state that the 50% overexpressed genes change the timing, degree, and rules for development. According to genetic central dogma, we know that the genes carried by chromosomes regulate our development by adjusting the expression of specific proteins. Having an excess amount of protein produced could cause the symptoms mentioned before. Then human’s course of development is disrupted which increases the patient’s risk of acquiring other diseases [5].

For the amplified developmental instability theory, the extra genes will trigger non-specific chromosomal disturbance which can disrupt homeostasis. Transcription from DNA to RNA is a complex process that requires many enzymes and molecules to work together in the right way. However, an extra chromosome makes it difficult for the transcription to proceed normally and drives our cells into a state of chaos [6].

To find the exact genes influenced by the extra chromosome, scientists analyzed the RNA produced by Down Syndrome and those produced by normal people. The difference in gene expression for some genes on chromosome 21 is obvious: 25 genes’ expression increased significantly. Moreover, around 85 genes on other chromosomes increased their expression while about 100s’ expression decreased. Studies relating to The Ts65Dn and the Ts1Cje region on orthologous genes from mice also proved the results of observation.

2.3. The symptoms of down syndrome

It is an easy thing to recognize a Down Syndrome patient due to their apparent physical features: They have flattened faces and a small head compared with normal people. To include more detail, people with Down syndrome also show short height, necks, and fingers. Smaller sizes were not shown for Down Syndrome infants they look smaller because of slower development after birth.

Down syndrome trouble parents more than just changing their children’s outlook. Intellectual disability is what makes it a “syndrome”. It impairs the patients’ cognitive skills and reduces memory. For instance, Down syndrome children master language later than the average. Learning is difficult for them and thus they have to have specially cared for [7].

Something about down syndrome is even worse: patients with Down syndrome have a high risk of getting Alzheimer’s disease. Alzheimer’s disease not only occurs frequently among patients but also develops at an unusually early age, the most time during their 40s. Alzheimer’s disease makes them worse in memory and recognition and can prevent them from communicating with families-because Alzheimer’s causes one to isolate him or herself from others, and become unwilling to talk.

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Alzheimer's disease is caused by the dysfunction of neurons in the human brain. In other words, neurons are damaged and even destroyed. There are two ways this can happen:

The formation of neurofibrillary tangles can cut the supply of nutrients to the neurons and eventually lead to their death. Neurofibrillary tangles are made of packed strands of protein tau which must be in lines to transport the nutrients. Being shaped in tangles makes them fail to perform their function.

In addition, the forming of beta-amyloid plaques can cut the signaling between neurons. Amyloid plaques are clusters of misfolded protein, when they accumulate to great extent, nerve cells can no longer transport signals to each other.

Down syndrome interferes with the process by making the accumulation of amyloid proteins easier than in normal conditions. The chromosome in which patients with Down Syndrome have an extra copy, chromosome 21, codes for a special gene that produces amyloid protein. The accelerated built up of amyloid protein damage the neurons of patients and eventually leads to Alzheimer's.

Due to bad gene regulation for Down Syndrome patients, 50% of them have congenital heart defects that put them at risk for early dementia, which is a symptom of Alzheimer's.

The last point is, that these patients often experience accelerated aging which makes them look older than they are. This can originate from the failed timing and patterns of development caused by trisomy 21 [8].

Scientific data provided by a former experiment pointed out something not expected: Some symptoms of Down syndrome are gender-related, and males are significantly at higher risk of having severe behavior problems compared with females (29.5% and 12.8%) [9].

2.4. Ways currently used to deal with down syndrome

It is a pity that the illness cannot be easily treated with a vaccine. For Down syndrome, the extra chromosome is unable to be changed since no technology now can accurately engineer special genes on a large scale without having too many negative effects on the patients. The only way to deal with Down syndrome is to prevent having babies born with it and protect those patients with medical and emotional aids. Technologies make it possible for parents to take an early diagnostic test and see if their babies are inclined to have Down Syndrome. Moreover, data have shown that the risk of having Down syndrome babies increase when the mother's age increase, so mothers may consider their ages if they are planning to have a child.

3. Nursing care of down syndrome

Since people with Down syndrome have intellectual disabilities, they are sometimes discriminated against by local communities. But people nowadays are trying to solve this problem: by offering support to patients' daily life and teaching them to live independently, A DDN (developmental disability nurse) can help them to fit into their local communities and enjoy mental safety [10].

The patients must be specially cared for, like having jobs with little pressure, being treated nicely by people around them, and offered opportunities to get married. However, being treated differently does not mean that people should regard them as strangers from their communities. Down Syndrome patients have dignity and they are part of our communities. As a result, nurses must realize that they should show respect to their patients' identities, and Down syndrome patients should be informed that they are as equal as others in rights given by the law. Feeling isolated from others will cause them to be inclined to have psychological problems and become dependent on a few people, which means being unwilling to get in touch with the outside world.

Nursing the patients involve teaching them to live themselves, so DDNs need to show the steps of daily work to the patients over and over again until they finally master them. It takes effort, and it works to help the patients feel better about themselves and generate motivation toward better lives.

In some areas on earth, people with Down syndrome are neglected and simply called "stupid". Some parents even abandon their children when they look like Down Syndrome kids. To change this situation, international organizations must take action to force local governments to reinforce laws to

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protect these patients' rights. The idea of protecting vulnerable groups like Down Syndrome patients are required to be popularized.

After all, with care given by trained nurses or NGOs, Down Syndrome patients' lives are still incomplete if their families do little in supporting them. Love within families is a strong power to make a patient happy, so family accompany is indispensable.

4. Conclusion

Down Syndrome is incurable. however, doctors and nurses make it controllable through hard work. Although there are ethical issues regarding whether or not to give birth to Down Syndrome babies, careful nursing that respects their own personalities enables them to enjoy a great life just like normal people. As a disease caused by the genetic disorder trisomy 21, current ways to deal with it is to take tests before babies are born and decide whether or not to give birth. Nowadays scientists believe Down Syndrome patients usually exhibit slow body development due to disrupted timing of development caused by extra genes. With a deeper understanding of the biochemistry bases of Down syndrome, we may one day figure out new procedures to protect this vulnerable minority group.

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