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Case Report

The Genetic Mutation of Human Cells, Somatic and Germs Cells Mutation in Human Body

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ABSTRACT

Substantial cell transformation can make hereditary fluctuation in a cell populace and can incite malignant growth and tumor when quality changes occurred at repressor quality in controlling cell cycles, for example, p53 quality. While germline cell change can cause hereditary ailment, for example, sickle cell iron deficiency, bosom malignant growth, thalassemia, Parkinson's just as deformity of biochemical pathway that impact sedate receptor collaboration, which has negative impact and lead to hospitalized of patient. The vast majority of reports referenced that point change, for example, a solitary base of nucleotide replacement (purine supplanted by purine or transversion (purine supplanted by pyrimidine or the other way around) that influenced hereditary infection just as antagonistic medication response that included hereditary components. Change that happened in germline cell would be acquired to the offspring, and these transformed qualities can spread in a populace through preparation process. Transformation that happen in coding casing of DNA district which of their demeanor are answerable for blend of explicit items could be ascent of hereditary infection, in light of the fact that the loss of quality capacity.

Keywords: Genetic mutation; Cells; Somatic; Germs; Human body

INTRODUCTION

A gene mutation is a lasting adjustment in the DNA arrangement that makes up a gene, to such an extent that the grouping varies based on what is found in the vast majority. Transformations extend in size; they can influence anyplace from a solitary DNA building square (base pair) to a huge fragment of a chromosome that incorporates numerous qualities [1].

A lot of factors caused mutation occurs in gene level especially for somatic cell mutation. Gene mutation (point mutation) can occur spontaneously that is caused by several of source or occurred error during DNA replication, chemical carcinogenic, virogenetic and ionic radiation. Similarly, methylation and transposable genetic element could give abnormal gene product with behave like gene mutation [2], but DNA methylation and transposable genetic element sometimes only provide temporary prevents of a normal gene expression that lead to change of phenotypic performance called as epigenetic effect [3]. Even recent report mentioned that epigenetic can lead to change of phenotypic performance called as epigenetic effect.

Epigenetic is a biological term, methylation is one the cause factor of epigenetic which usually occur methylation at Cytosine become methyl cytosine while lysine 9 of histon H3 methylation influenced assembly of chromatin structure [4].

Gene mutation is source of genetic variance and genetic polymorphism, these gene mutations can arise several possibilities i.e. genetic diseases, receptor mutation, and Adverse Drug Reaction (ADR) in human [5]. This adverse drug reaction in human body can be harmful for health and the life, most of cases are reported that mutation type is point mutation that brings about defective some P450 enzymes, examples: CYP2D6 and CYP2C9 gene mutation [2]. Both of thegene mutation of drug-metabolizing enzymes and another defective protein function in human can make shorter human life span. A lot examples, gene mutation influences biochemical pathway that caused metabolite disorder such as Parkinson's, Alzheimer, x, thalassemia, and hemophilia. Parkinson's gene mutation has been investigated from Parkinson's family history [1], the gene called Park 8 and make protein named "dardarin" for the Basque word for tremor, which indicate a symptom of the progressive and fatal nerve disease. Other genetic disorders are Hemoglobinopathies and thalassemia, while most of cases were reported that mutation in coding frame of these gene occurred substitution of amino acid that arrange a polypeptide chain of hemoglobin [6]. Hemoglobin (Hb) is a four chain (tetrameric) oxygencarryingprotein ofred blood cell (erythrocytes). Two major globin genes product, i.e., .α and β chain, two symbols for α- globin genes are HBA1 and HBA2, whereas symbol for β globin gene is HBB [7]. To make easy understanding, sometimes researcher provides symbols for hemoglobin gene mutation such as Hbs (HBB, Glu6Val) and HbMiyano (HBA1, Thr41Ser). The meanings of the symbols are: Hbs, hemoglobin associated with sickle cell anemia; whereas Glu6Val substitution glutamate to Valine. Hundreds variant of allele were detected from patient which has sickle cell anemia for examples it found that 189 allelic variant from HBA1, 34 for HBA2, 276 for HBB. This study was to determine the factors affecting genetic mutation of cells in human body, somatic and germs cells mutation types.

Somatic and Germ Cells Mutation

Eukaryotic organism consisted of billion of cells included both reproductive and somatic cells. The mature eukaryotic organism, there are two cells types i.e. Somatic cells (diploid) and germ cells or reproductive cell (haploid). Reproductive cells are produced by meiosis cell division derived from [8]. Diploid cell in mammals, somatic cell mutation cannot be inherited to progeny, whereas mutation take place in germ cell lines will be transmitted to progeny during fertilization process. Somatic cellsare diploid and they have one set of chromosomes, they are descended half from mother and half from father, while reproductive cells have half set of chromosomes called haploid [9]. A lot of reports concerning with Eukaryotic organism consisted of billion of cells included both reproductive and somatic cells. The mature eukaryotic organism, there are two cells types i.e. Somatic cells (diploid) and germ cells or reproductive cell (haploid). Reproductive cells are produced by meiosis cell division derived from. Diploid cell in mammals, somatic cell mutation cannot be inherited to progeny, whereas mutation take place in germ cell lines will be transmitted to progeny during fertilization process. Somatic cells are diploid and they have one set of chromosomes, they are descended half from mother and half from father, while reproductive cells have half set of chromosomes called haploid [4]. A lot of reports concerning with somatic cell mutation that caused tumor or cancer in human such as prostate cancer, as well as p53 gene mutation result in breast tumor or cancer. Somatic loss of BRCA1 and p53 that cause repressor gene function such as p53 in active and lead to BRCA1 and BRCA2 tumor, BRCA1 transcriptionally regulate genes involved in breast tumorigenesis [10].

Somatic cell mutation in plant can be inherited into progeny because functional cells are able to differentiate into mature plant. One of example for somatic cell mutation is transgenic plant (insert mutation) [11]. Insertion of foreign gene into single somatic cell

(vegetative cell) that is able to provide totipotence into mature plants that will give reproductive cells (ovum and pollen) that bring inserted gene and it is transmitted into next generation.

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Genetic Basis of Receptor and Mutation

Receptors are macromolecule (protein) involve in chemical signaling between intra and extra cellular. They are located on the cell surface and trans membrane examples: receptor of acetylcholine, GABA, growth factors, and insulin receptor, whereas nuclear receptor present in cytoplasm such as steroid hormone receptor, thyroid hormone receptors and androgen receptor. According to Laudat, et al. [12] nuclear receptor derived from common progeny based on the DNA binding C domain, which can be grouped into three sub families i.e. thyroid hormone and retinoic receptor, orphan receptor, and steroid hormone receptors. Based on the types and function of receptor, it can be divided four types i.e. Ion channels (Ligand-gated ion channels), G-coupled receptors, enzymes-linked receptors, nuclear hormone receptors. Receptor can be activated by physiological process in the cells and ligand binding site such as drugs, hormone and neurotransmitters. Molecular biology development techniques aided to study drug-receptor interaction at molecular level. Receptors are macromolecule (protein) involve in chemical signaling between intra and extra cellular. They are located on the cell surface and trans membrane examples: receptor of acetylcholine, GABA, growth factors, and insulin receptor, whereas nuclear receptor present in cytoplasm such as Steroid hormone receptor, thyroid hormone receptors and androgen receptor Receptors are macromolecules (protein) that involved in chemical signaling between intra and extra cellular. They are located on the cell surface and transmembrane examples: receptor of acetylcholine, GABA, growth factors, and insulin receptor, whereas, nuclear receptor present in cytoplasm such as steroid hormone receptor, thyroid hormone receptors and androgen receptor.

CONCLUSION

Mutation can alter biochemical pathway of drugs metabolism because a lot of gene products of CYP450 related to drugs metabolism which involve pharmacokinetic and pharmacodynamics genes function and can lead to adverse drug reaction in human. Similar phenomena was founded on genes mutation that involved drug receptor interaction that influenced drug transport into cell target, it can rapid or slow, hence consideration about genetic factors that influenced drug metabolism is necessary.

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