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The Study of Bioinformatics Application for Von Hippel -Lindau Disease

RABINDRA KUMAR MISHRA[1]*, PRANJALI POTDAR [2], SHILPA WAKHARE[3]GAJANAN SONAWANE [4,] PADMALAYA MISHRA[5]

[1]*Department of Basic Science and humanities, GIET University, Gunupur, Rayagada, Odisha, India, 765022*
[2345] *Department of Biotechnology, GIET University,Gunupur, Rayagada,Odisha, India, 765022*

Abstract:
This article gives an overview of neurodegenerative illnesses, a genetic defect that causes Von Hippel –Lindau disease. This article also provides the concept of various forms of tumors in their body. In this study, Genetic testing is executed to check for the anomalistic gene in own circle of relatives A case history of von Hippel-Lindau sickness and a minimum of one characteristic neoplasm within the eyes, brain, spine, adrenal glands, kidneys, or duct gland are present. Therefore, developing drug molecules could help researchers compare early and late-stage forms of the disease. This could offer new pharmacological targets for treatment as well as processes particular to late-onset von Hippel -Lindau disease.

Keywords: *Lindau Syndrome,central nervous system,tumor,angiogenesis*

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I. Introduction

Von Hippel-Lindau Disease is autosomal dominant inheritance also known as VHL.Angiomatosis retinae, and Familial Cerebello-Retinal Angioma .It is a very uncommon genetic condition characterized by the growth of tumors and cysts (fluid-filled sacs) in a variety of bodily organs, including the skin, eyes, and central nervous system (CNS).VHL disease is caused due to genetic alternation (mutation iteration of the alleles of the VHL gene situated on third chromosomes'(1). Through its tumors the VHL protein, gene acts as a key regulator of cellular hypoxia signaling (pVHLIncreased levels of growth factors like platelet-derived growth factor, transforming growth factor-alpha, and vascular endothelial factor are inadvertently caused by pVHL. Regulate factor-alpha complex does not occur in the case of a non-functioning gene, such as in VHL disease. As a result, the concentrations of various growth factors increase, increasing the efficacy of angiogenesis and tumor development.VHL affected people having various types of tumors in their body i.e. tumors in the brain, inner ear, spinal cord, kidneys, eyes, pancreas and adrenal glands, reproductive tract, brand east(2). Some tumors errand cancerous (malignant) and grow rapidly and spread throughout the body and othershavenoncancerous (noncancerousis a rare genetic disorder and it affects 1 person out of 36,000 people.

Traits of VHL:

- Migraine
- Hearing loss or ringing in the ears (tinnitus)
- High blood pressure
- Loss of balance
- Loss of muscle strength or coordination
- Vomiting
- Vision problems

Clinical Features:

Hallmark depending upon tdependon of the tumor in organs

1. Hemangioblastomas-tumors associated with brain and spinal cord

- Migraine
- Weakness
- Ataxia
- Nausea
- Vomiting

- Retinal-blindness

2. Pheochromocytomas-Tumors found in adrenal glands

- An acute increase in blood acupressure
- Migraine
- Fear strikes
- Profuse sweating

3. Renal Cell Carcinoma-Tumors present in kidney

- hematuria
- lower back pain
- anemia
- loss of appetite
- weight loss
- Fatigue
- fever
- Development of a lump on one side.

4. Endolymphatic Sac Tumors-tumors formed the in inner ear i.e.benigthen in Nature

- Hearing loss
- Tinnitus (ringing/buzzing in the ears)
- Problems with maintaining balance while standing and/or walking.

5. PancreaticNeuroendocrinein Tumors-tumors associated with islet cell Tumor

- Hypoglycemia (low blood sugar)
- Stomach ulcers
- Gallstones
- Severe diarrhea.

Diagnosis of VHL:

1. Imagine g tests
2.Eye examination
3. Genetic testing

Doctors perform a physical examination and verify for von Hippel-Lindau disease in any family and friends(3).Various tests are conducted to look for tumors and other abnormalities if findings point to the disorder:the spine and brain using magnetic resonance imaging (MRI) or computed tomography (CT) an ophthalmoscopy-based eye examination abdominal MRI, CT, or ultrasonography audio tests a blood test.

A DIAGNOSIS OF VON HIPPEL-LINDAU DISEASE IS MADE WHEN ANY OF THE FOLLOWING CONDITIONS EXIST:

Doctors examine patients who have two or more typical von Hippel-Lindau disease tumors but no known von Hippel-Lindau family history. Once one tumor is discovered, they search for more.To check for the anomalistic gene in one's own family tree, genetic testing is done(4). A case history of von Hippel-Lindau disease and at least one distinctive neoplasm in the kidneys, adrenal glands, eyes, brain, spine, or duct gland. Molecular cistrontic testing is used by doctors to identify the abnormal VHL gene and confirm the diagnosis if they are still unsure of the condition(5). Genetic testing is carried out to check for abnormal citron in family members if the abnormal VHL gene is found in one particular person(6)(7).

Treatment:

Person ring from VHL either opts to get tumoredthrough surgery or sometimes radioradiotherapy our removal is not possible before it getsexgetsciating radiation are targeted on tumor insteadOptical device medical aid such laser as therapy or utmost cold application is employed forangias of the retina(8,9).

Belzutifan drug which is approved is quite frequently used for adults to reduce neoplasm in various organ liorgansmors in the kidney, brain, or spine, or tumors in the pancreas that doesnot reseed to be immediately surgically removed(10,11).This medication reduces tumor size and halts tumor growth. It may be used up until the disease gets worse or the side effects get too bad. severe who have adrenal gland tumors may also require medication to regulate their blood pressure. Advanced kidney cancer patients might also receive other drug therapies(12–15).

Related drugs:
Drugs
utifan
Tivozanib
Osilodrostat
Indium In 111 pentetreotide
Serpin B5
Plasminogen
Neocarzinostatin
Rhein
Pazopanib Hydrochloride (clinical trial)
Fucoxanthin
Abivertinib
E3 ubiquitin-protein ligase TRIM13

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II. CONCLUSION:

This article gives an overview of the causes of Von Hippel –Lindau disease. , the levels of various growth factors rise, allowing for increased blood vessel growth (angiogenesis) and tumor formation.

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