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Genetic Effect and Environmental Analysis of Various Genetic Diseases.

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Review Article

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ABSTRACT

Genetic and environmental factors are the main key factor in the mechanism of various diseases. Previous studies have shown that environmental factors can also be the main cause in the pathogenesis of most diseases. The review details a brief overview discussing the environmental and genetic factors influencing the diseases. This review also emphasizes the findings categorizing the common diseases on the basis of genetic profiles, disease diagnosis, and treatment based on the genetic influence.

INTRODUCTION

Genetic diseases

Genetic syndrome is basically genetic problem which is caused due to abnormality in individual's total genetic content. Abnormality can be minor i.e single base mutation or major i.e involve in the entire chromosome. Some diseases like cancer occur due to abnormality in genes or chromosomes or can be caused/influenced by environmental factors. If a syndrome is caused by the combination of both genetic and environmental factor and then genetical origin of the diseases can be referred as having multifactorial system. Various diseases including cancers along with a plethora of other diseases, are concluded to be a result of environmental triggers.

Genetic factors of diseases

Diseases to be discussed are:

- Sickle cell disease
- Attention deficit hyperactivity disorder
- Chronic Neutrophilic leukemia
- Oral-facial-digital syndrome
- Arterial Thrombosis
- Insulin dependent diabetes mellitus
- Huntington's disease
- Breast Cancer

Sickle cell disease is also known as sickle cell anaemia comes under hereditary blood disorder, that affects haemoglobin. The pathology of sickle cell sickness, caused by sickling appearance. In this disorder the body makes sickle shaped red blood cells. In deoxy state hemoglobin S molecules aggregate into long polymers that are aligned for the formation of liquid crystals known as "tactoids". In the

presence of tactoids the red cell is transformed into the characteristic sickle shaped cell is the reason for the particle disease name. There is increase in blood viscosity formed with small vessel stasis and obstruction, which leads to organ damage. Furthermore patients having sickle cell disease have a propensity to bacterial infection owing to combination of many disorder [1]. Apart from that the effect of environmental factors, the occurrence of non-communicable diseases are determined by non-genetic and genetic developmental factors [2]. Attention deficit hyperactivity disorder (ADHD) is one of the most common disorder. It is commonly studied and diagnosed psychiatric disorder in children, affecting about 3 to 5% of children worldwide [3]. Chronic neutrophilic leukemia (CNL) is a rare myeloproliferative disorder characterized by persistent mature neutrophilia in peripheral blood. Because the disease entity is rare, and because it is typically a diagnosis of exclusion, it is important for pathologist to be familiar with CNL when approaching the patient with a myeloproliferative clinical picture [4]. Preoperative Iron deficiency anaemia (IDA) is common with poor postoperative outcomes. Standard treatment includes oral iron supplementation and allogenic blood transfusion. This trial demonstrates that intravenous iron is both a feasible and effective treatment for IDA in anemic colorectal cancer patients [5-15]. Arterial thrombosis is a blood clot inside blood vessel. It blocks the flow of blood in the circulatory system. Huntington's disease is a neurodegenerative disorder which affects muscle coordination leads to mental symptoms. It first affects the brain, with gradual loss of control of memory and mental capability. Breast cancer is a type of cancer that develops from cell of breast or may be a lump in breast. One study shows the possibility of using ANXA7 as both a clinically relevant indicator of disease progression and a prognostic biomarker for survival in the patients with triple negative breast cancer. ANXA7 might serve as promising target for triple negative breast cancer therapy. The gene for Huntington disease on the fourth chromosome has been characterized [16-38]. Gastrointestinal diseases are functional disorders, as being dominant in women, but others, as being more common in men. The pathogeneses of gastrointestinal diseases are still unclear but steroid sex hormones play important role in the pathogenesis of several gastrointestinal diseases [39-43]. Type 1 diabetes mainly known as insulin dependent diabetes mellitus (IDDM) is documented as an autoimmune disease in which pancreatic islet cells are progressively destroyed. The highest degree of enzyme specificity and fluorescence sensitivity arrows that enzyme inhibition assay is simpler, accurate and sensitive for screening a large number of serum samples) [44-47]. Schizophrenia is a psychiatric disorder characterized by abnormalities including elusions, conceptual disorganization, cognitive impairment, and hallucinations. However, investigations are required to illuminate the enlarge frequency of the D allele among schizophrenia patients [48]. Irritable bowel syndrome (IBS) and inflammatory bowel disease (IBD) damages the tissue of the small bowl and large bowel through the process of inflammation. Gastrointestinal diseases are recognized, as being dominant in women, but others are more common in men (Anal fistulae). The observations elevate concerns about the involvement of sex steroid hormones in the pathogeneses of these diseases [49]. This prospective study assessed potential efficacy and toxicity of low-dose granulocyte macrophage colony-stimulating factor (GM-CSF), interleukin 2 (IL-2) and interferon alpha (IFN) postoperatively in patients with highrisk renal cell carcinoma (RCC) [50]. Prostate cancer (PC) is a form of cancer that starts in the prostate gland. Studies suggest that the prostate cancer incidence increased in Asian population in the last few years [51-55]. In HIV-1 infection, some genetic factors may be important as for example the gene variants, encode chemokine receptors: CCR5, CCR2 and CXCR4 - SDF-1 ligand receptor. The finding might be helpful for epidemiological researches of HIV infections and other diseases like diabetes [56]. Asthma and chronic obstructive pulmonary disease (COPD) show resemblance and significant differences. This is specified that asthma and COPD have mutual genetic and environmental risk factors which eventually lead to clinical disease depending on the timing and type of environmental exposures. The genetic predisposition to certain pathways may further help to define the development of COPD. At the end this may lead to stratification of patients by their genetic make-up and open new therapeutic prospects. Juvenile Myelomonocytic Leukemia (JMML) is a serious chronic leukemia which affects young children. Myeloproliferative/myelodysplastic (MPD/MDS) hematopoietic disorder caused by progressive bone marrow failure. Metabolic syndrome is represented as a cluster of conditions, increased blood pressure multiple risk factors, counting central obesity, hypertension and weaken glucose tolerance, that enhances cardiovascular disease morbidity and mortality [57-59]. Ovarian cancer is the fourth leading cause of cancer-related death in women and also the leading cause of gynecologic cancer death. More specifically, epithelial ovarian cancer (EOC) is characterized by few symptoms, display at an advanced

stage and poor survival. Many cases around 190,000 new cases of epithelial ovarian cancer (EOC) each year worldwide and this malignancy represents the leading cause of death from gynecological cancers. Possibility of using ANXA7 as both a clinically relevant indicator of disease progression and a prognostic biomarker for survival in the patients with ovarian cancer [60]. Chronic infantile neurological cutaneous and articular syndrome (CINCA), also known as neonatal-onset multisystem inflammatory disease (NOMID), is dominantly-inherited systemic auto inflammatory disease. Some Neuroendocrine tumours (NET) occur in hereditary-familial neoplastic syndromes such as MEN (multiple endocrine neoplasias) or neuroectodermic dysplasias while others arise as solitary-isolated neurofibroma [61]. HNF1B nephropathy is typically responsible for bilateral renal cystic hypodysplasia in childhood one findings have provided data that are useful for recognition and diagnosis of HNF1B disease in adulthood and might help in renal management and genetic counseling [62,63]. Genetic defects of platelet function give rise to mucocutaneous bleeding of varying severity because platelets fail to fulfill their haemostatic role after injury in vessel [64]. Mendelian inheritance has been demonstrated for some disorders while others are associated with mutations and polymorphisms in susceptibility genes [65]. If stable gene transfer is needed, non-integrating vector systems may be combined with transposon-or phage integrase-based systems or future site-specific systems to achieve integration into the host B cell genome [66]. First time a de novo chromosomal abnormality which produced the phenotype of a female with primary ovarian failure and subsequent osteopenia in early adult life [67]. Mutations in several multidrug resistance proteins (MRPs) are associated with human genetic disorders [68]. Dravet syndrome, also called severe myoclonic epilepsy of infancy (SMEI), is a severe form of epilepsy. It appears during the first year of life with frequent febrile seizures. The Dravet syndrome also known as Severe Myoclonic Epilepsy of Infancy (SMEI) begins in the first year of life is a rare and catastrophic form of intractable epilepsy [69]. Fanconi Anemia (FA) is a genetic disease in cancer research, and the most reported of the rare inherited bone marrow failure syndrome [70]. Genetic, hormonal and life-style related factors determine SHBG levels and low sex hormone-binding globulin levels are a known risk factor for the development of the metabolic syndrome, diabetes and cardiovascular diseases [71].

Environmental factors influencing diseases in different ways

During the past few years, experimental evidence has emerged to suggest that environmental factors may influence cellular proliferation attrition in an organ-specific manner. These cases, and in disorders there is no genetic background but environmental factors appear to influence and/ or cause disease onset, progression, and outcome [72]. Traditional risk factors for breast cancer explain only a fraction of cases. Source for tendency in breast cancer incidence are not fully comprehended. Breast cancer incidence and mortality rates decrease with environmental conditions that promote Vitamin D synthesis in human skin including lower latitude and higher personal exposure to sunlight [66]. Environmental factors are threats to health and managing them is public environmental health. They include [73].

- Environmental conditions support disease vectors (endemic and exotic vectors)
- Invasive biota and their hosts and vectors
- Environmental interference: floods, droughts, storms, earthquakes, volcanoes, fires
- Air condition: pollen and pollution leading to respiratory diseases or cancers
- Water nature: biotic and abiotic contaminants; integrity of water transport and treatment infrastructure
- Monitoring and management of agricultural, industrial outflows to the environment

Gene therapy and other diagnosis factors in cure of diseases

In quality treatment the incorporation methodology of the viral DNA genome into the host cell genome is an essential venture for infection joining. Few years ago, distribution of retrovirus was accepted to be arbitrary and the shot of incidentally actuating a quality was viewed. Tumorigenesis connected to a few studies in quality treatment is suspected to be created by insertion process. It lies on whether the provirus incorporates into or in the region of qualities typical transcription can be upgraded or disturbed in this way impelling oncogenic changes. It is called as insertional mutagenesis. This researches whether territory over the genome could be supported by retrovirus mix is a vital angle in quality treatment. These region are called "Regular Integration Sites" (CIS) or "hotspots" [74]. Recombinant adeno-related infection (rAAV)-based quality treatment speaks to a promising methodology for the treatment of heart muscle ailments, where as the sub-atomic systems that head-on rAAV transduction stay uncertain. It is reasoned that, calcium-subordinate pathway directs rAAV vector transduction at various stages that may incorporate vector transformation, assembly, and translation action. Managing this pathway through β -adrenergic flagging improves rAAV-interceded quality conveyance to cardiomyocytes, and it is significant when considering helpful methodologies for heart muscle infections [75]. Liposomes are perceived as imperative vehicles for cytotoxic medications in light of the fact that they can shield the medications from degeneration in movement, consequently securing fit cells, tissues from disclosure to deadly medication measurements. Liposomes have been embraced as tumor-particular and compelling transporters of cytotoxic medications. At the same time, they are not missing of noteworthy issues including untimely decimation to bring about lethality to solid tissues and in the other great, unwantedly long haul dependability to avoid powerful conveyance to the tumor cells [76]. Plasma B-sort natriuretic peptide (BNP) qualities have been assessed as indicators of result and are useful in determination of helpful choices in patients with both intense and incessant heart disappointment [77]. Early and expanded measures of plasma have been connected with enhanced survival in the wake of entering. Anyway, no studies including blaze patients exhibit the impacts of Intraoperative plasma organization on postoperative revival need. This examination concentrated on perioperative transfusion proportions and the part of untimely, forceful plasma organization in contemporary smolder midpoint [78]. Not by weakening itself but rather likewise by more confused correspondence, restorative liquids utilized as a part of serious consideration can have significant impacts on focus. Point of some study was to screen the degree of dilutive coagulopathy incited by the most widely recognized helpful liquids and to endeavor standardization of haemostasis by fibrinogen expansion [79]. At present various nations including United States screen all gifts for hostile to HBc which is not required in some different nations, for example, Iran. By the by it has been contended that the avoidance of hostile to HBc positive contributors is unrealistic in nations where HBV contamination is predominant and more than 20 percent of the populaces are sure for against HBc [80]. There have been late discoveries on cobalamin insufficiencies because of nourishment cobalamin malabsorption or nondissociation of vitamin B12 from its bearer proteins disorder [81]. The quality platelets concentrate assumes a vital part in transfusion treatment [82]. It has been given that the confirmation of the articulation profile of GR proteins in the human myometrium and have shown a pregnancy related downregulation. Further research is expected to comprehend the physiological noteworthiness of this perception and to clear up the part of glucocorticoids and GR in pregnancy support and the onset of work [83]. Metabonomics is formally characterized as "the quantitative estimation of the multi-parametric metabolic reaction of living frameworks to pathophysiological jolts or hereditary change". This methodology is reciprocal to proteomics and genomics and is appropriate to an extensive variety of issues in various biomedical examination ranges [84]. Advancement of nanoparticles as specialists for focused on location of growth cells through imaging has been an energizing range of examination lately. Exact focusing on is of discriminating significance especially when these specialists are likewise utilized for moving restorative atoms to treat particular tumors or tumor. Attractive reverberation imaging (MRI) remains an alluring imaging stage because of its high spatial determination [85]. Atomic tumor profiling has potential significance in distinguishing the tissue of root in patients with disease of obscure essential [86]. There have been a few transformations distinguished in the apoB-100 quality prompting untimely truncation of protein blend or to amino corrosive substitution inside the protein such changes can impact the digestion system of plasma lipoprotein and may accordingly be vital in the advancement of hyperlipidemia and coronary illness [87]. Beforehand, It has been accounted for that ordinary ovaries need neutrophil gelatinase related lipocalin (NGAL) articulation and that NGAL statement happens in kindhearted tumors

and increments in ahead of schedule evaluation ovarian tumors [88]. Constant subcutaneous insulin mixture (CSII) treatment is generally acknowledged for fragile sort 1 diabetes, since it has the advantage of less recurrence of hypoglycemia and better administration of the sunrise wonder contrasted with different day by day insulin infusion (MDI) treatment [89]. Near studies in the middle of African and European populaces recommend that aggregate lymphocyte check (TLC), including CD4+ number, is liable to fluctuate essentially by ethnicity, in both, solid and HIV-contaminated Individuals [90]. Common executioner (NK) cell capacity was researched in Malaysian HIV patients starting antiretroviral treatment (ART) with cutting edge immunodeficiency. A few patients experienced resistant reclamation illness (IRD) displaying as intensifications of previous diseases. Whilst most IRD are credited to interferon-gamma created by T-cells, NK cells might likewise contribute [91]. As of late specific consideration has been attracted to the impact of antiretroviral (ARV) treatments on the rate of genuine non-AIDS occasions (SNAEs), including cardiovascular infection (CVD), end-stage renal ailment, liver disappointment and breaks specifically noteworthy is the impact of individual ARVs or ARV classes on these occasions [92]. Insusceptible Recovery Inflammatory Syndrome (IRIS) is described by a confusing disintegration of clinical status after launch of Anti- Retroviral Therapy (ART), regardless of enhanced resistant capacity. It is brought about by provocative reaction against the irresistible antigen [93]. It takes after an already asymptomatic man who gave normal, non-particular manifestations and was diagnosed with an uncommon confusion of non-typhi Salmonella contamination. This serves as an unmistakable suggestion to all clinicians to have a low limit for testing for HIV and highlights the need to present a withdraw framework for HIV testing in all intense medicinal services settings with a high commonness of HIV contamination [94]. Attendant antiretroviral treatment (ART) can be a component prompting a lower adequacy of pegylated interferon (peg-IFN) in addition to ribavirin (RBV) in human immunodeficiency infection (HIV)/hepatitis C infection (HCV)- confected patients. In this study, HIV/HCV-confected patients who were treated with a three-medication regimen including TDF in addition to 3TC or FTC alongside NVP reacted better to peg-IFN in addition to RBV than the individuals who took LPV/r, a finding that has not been beforehand reported [95]. Indeed, even with mix antiretroviral treatment (cART) regimen, the solidness of HIV control is constrained by numerous components (adherence to treatment, drug harmfulness, bioavailability, among the most essential. At the point when rescue treatment is viewed as, better result is normal if antiretroviral regimen incorporates a class to which the patient has not been uncovered beforehand. Thusly classes of antiretroviral medications coordinated at targets other than converse transcriptase or protease are of potential extraordinary interest [96]. Studies planned to evaluate the impacts of medications in patients with safe disappointment, and in addition on the frequency on AIDS and non-AIDS occasions, liver fibrosis and atherosclerosis movement are unmistakably required [97]. Histoplasma is a dimorphic pathogenic growth which causes human disease around the world, for the most part in central nations [98]. Irresistible creatures, probably infections, have long been a suspect for setting off the immune system reaction in individuals hereditarily defenseless to MS. Albeit numerous irresistible microorganisms have been researched, no specific living being has risen as a demonstrated trigger The AIDS infection is a neurotropic infection and CNS inclusion as the displaying dissention is seen in more or less ten percent of instances of HIV contamination [99]. Osteopontin (OPN) is a discharged phosphoprotein which assumes a discriminating part in metastasis of colon, liver, and bosom diseases [100]. Epigenetic regulation of quality statement, through covalent change of histones, is a key methodology controlling development and improvement. In like manner, the translation variables directing these methodologies are vital focuses of hereditary infections [101].

CONCLUSION AND FUTURE DIRECTIONS

This survey is intended to concentrate on the impact of host and hereditary considers the pathogenesis of malady and its determination. These studies must be repeated in distinctive populaces for deciphering the instrument of pathogenesis of the infection. Future studies ought to investigate the instrument of quality modification focusing on the ailment movement from sound state. Regardless of late advances in the comprehension of the pathophysiology of human maladies obtained aplastic paleness and idiopathic aspiratory fibrosis, the conceivable reasons for these infections stay perplexing. Various studies have archived ecological elements.

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