

## Retinitis Pigmentosa (RP) at a Glance

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### ABSTRACT

Retinitis Pigmentosa, abbreviated as RP, is a global eye retina disorder. In this mini review paper, my focus just only its basic causes, prevalence, symptom, and factors and also discuss retina syndrome related to this disease, to provide a basic knowledge for beginners and readers to understand about what is retinitis pigmentosa disease.

**Keywords:** Retinitis pigmentosa; Causes; Symptoms; Factors; Syndromic retinitis pigmentosa

### INTRODUCTION

Retinitis Pigmentosa (RP) is a group of disorders which produce the repeated degeneration of the cells of retina. This degeneration normally starts from the mid-periphery then moves toward the macula and fovea [1]. RP is the most general hereditary retinal deterioration in the world [1]. "Retinitis" word is a misnomer as the soreness of retina is not imparting the major contribution to pathophysiology of the disease [2].

### PREVALENCE

The global occurrence of the retinitis pigmentosa is 1 in 4000 persons while in case of worldwide more than 100 million individual are expected with retinitis pigmentosa [3]. 3.4 million individuals of USA are affected by the damage of eyesight. It is estimated that number of affected people will be more in future because of the aging of the residents of modern countries [4,5]. High frequency of disease retinitis pigmentosa was observed in the Islands of Faroe. The notifiable frequency of retinitis pigmentosa differ from 1/4000-1/2500 [6]. Almost 1.5 million people in the world are involved in the inherited retinopathy which is the general form of retinitis pigmentosa [7]. In case of retinitis pigmentosa ratio of diseased people is almost 1 in 4000 in the common people [8-10].

### SYMPTOMS

Retinitis pigmentosa is genetic disease caused by damage of retina and gradual degeneration of rod and cone cells and pigments of retina start to accumulate on the fundus examination. Rod-cone dysfunction is general form where the night blindness is the initial

sign and the continuous damage in the line of sight which finally lead to blindness after many years [11]. In the beginning rod cells start to damage then the cones cells also start to deteriorate which produces the unexpected result. Nyctalopia and gradual restriction in the field of vision is the result of damage of rod cells [12]. The disorder retinitis pigmentosa route is separated into different stages. Night blindness is the most important symptom at the initial stage. At the first year of age this symptom starts to appear. Mild blindness is also the symptom of RP which is normally unnoticed by the affected people but it might be considered in teen age then it shows the flaw of the peripheral field of vision in the faint light. In the presence of day light these defects do not exist [13]. In the second stage the night blindness disorder becomes clear then causes more problems during the night and in the evening walk because affected person is susceptible to the damage of the peripheral vision in the daylight. Due to this they could not see the people on the footpath or cars coming from the side. In handshaking they could not shake hand properly because they could not see the hand position properly. As a result affected people do not feel easy in the unknown places and night driving specially. They feel the difficulties in reading due to the improper light. This is due to loss of visual acuity and to some extant muscular participation. In the last stage affected members cannot walk independently due to the damage of the peripheral vision but it retains the some degree of the visual field near the fixation point. They face hurdle in reading so they need magnifying glass. In this stage the condition of photophobia is more severe [13].

### CAUSES OF RP

Retina is a coating of thin tissues in the reverse side of eye

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which is the main source of vision and recognition of beam of light [14]. Originator cells of the retina in case of vertebrate's cells are the derivative of the neuroectoderm. They are divided into six types of the neuronal cell which are organized into seven layers [15]. In these layers one is the Ganglion Cell Layer (GLC), IPL and INL (inner plexiform and nuclear layers), PCL (Photoreceptor Cell Layer), RPE (retinal pigment epithelium). Ganglion cells layer consist of ganglion cells that spread the visual signal to optic nerve. Photoreceptor cells layer consists of external segment of the photoreceptors which are the basically light sensitive neurons of the retina. nuclei of rod and cone cells and their synaptic connections are present in ONL (outer nuclear layer) and OPL (Outer Plexiform Layer). The Retinal Pigment Epithelium (RPE) is the last layer which does not have neuronal cells but motivates the preservation and development of PCL (Photoreceptor Cell Layer) and minimizing its effect by absorption of the extra amount of light [16]. The neurogenesis of these layers is organized by cellular factors and genetics of host. Mutation is distressing these layers and some other genes which are accountable for different different disease of retina and blindness in human [17]. Unusually RP is considered as heterogeneous. There are various forms of heterogeneity such as allelic heterogeneity, genetic heterogeneity and clinical heterogeneity. When various genes become source of disease of same phenotype it is due to genetic heterogeneity. When special mutation occurs in the same gene which produces diverse disease it shows phenotypic heterogeneity eg. mutation in rhodopsin become the source of Autosomal dominant RP whereas a few mutations in rhodopsin produce recessive RP condition. While in some cases various diseases producing mutations in each gene then it show the allelic heterogeneity. In case of clinical heterogeneity similar mutation in various persons which show unlike results although they belong to same family. Two types of retinitis pigmentosa are observed, one is non Syndromic RP while other is the syndromic form. Almost mutation identified in 56 genes becomes the source of non Syndromic RP. Almost 23 mutated genes show Autosomal-dominant retinitis pigmentosa, 36 mutated genes show recessive RP condition and 3 mutated genes account for X-linked RP. In case of systematic form mutation observed in 12 genes causes the usher syndrome while mutation in seventeen genes show the BBS (Bardiet-Biedl Syndrome) [18].

## FACTORS AFFECTING THE RETINITIS PIGMENTOSA

Genetics and environmental both factors increase the severity of the neurodegenerative disorders in the human.

### Environmental factors

Environmental factors have major contribution in the damage of retina in case of RP.

### Light exposure

In the start some changes in the tissues of retina were observed at once after the exposure of light. It was observed that dose - response relationship between the some changes in retina tissue

and the exposure of light. Large amount of light can produce the rapid loss of the neurons which causes the complete damage of photoreceptors cells less than four weeks [19]. Vitamin A can save the photoreceptors from the trophic effect. Similarly vitamin E can also protect the photoreceptors from the antioxidant effects. Supplementation of vitamin A the dose is 15000 units per day showed the undesirable effects [20].

### Genetics factor

Most general way of classification of RP based on the pattern of inheritance and involvement of the photoreceptor cells [1,2]. There are two forms of RP, one is primary and other is syndromic form. In case of primary RP disease the affected area is only limited to the eye organ. Different mode of inheritance in case of primary RP are given. Autosomal dominant form of RP is not generally considered the sever condition and its occurrence start after the age of 50. Sometime condition become serious. Autosomal recessive RP normally start from the first ten years. It is also considered the mild form sequentially to thrash out the genetics of isolated retinitis pigmentosa. In some sequence autosomal dominant inheritance is derived only those condition when vertical transmission of RP is characterized in three generation of the same family [8]. According to some other the two generation participation is enough due to the low penetrance of the autosomal dominant retinitis pigmentosa [21-23]. Autosomal dominant forms of RP is normally identified as minimal disease which progress slowly and also retain the central vision in the period of ten years or more [24]. Although this inherited kind of RP has a spectrum of phenotypes from the occurrence of the disease at early stage and then become severe, progress slowly at the late stage which is restricted to the damage of rod and cone cell [25]. A few pedigree shows the minimum penetrance for these genes [26]. In case of autosomal recessive (RP) where the affected individual are identified with normal parents and multiple diseased siblings [27]. It is observed that poor dark adaptation at the juvenile onset progress fastly as compared to autosomal dominant retinitis pigmentosa. In case of autosomal RP more than one non-syndromic forms are available [9]. Uncommon form of RP is x-linked form but it is characterized as serious form. In this case the generally affected males face the problem of visual impairment at the age of mid-30 years to start of 40 years. Rare pattern of inheritance is observed in case of digenic and mitochondrial DNA. In case of digenic RP mutated genes found on the two various chromosomes in the same person. The interface of two genes produce the RP. some cases which are connected with real-time heterozygous mutations in the ROM1 while heterozygous mutation is observed in the periphery or sds genes [28]. In case of mitochondria it was observed that they have their own DNA, It is genetically transmitted from mother. Damage in the mitochondrial DNA causes the many disease. Due to the complications in the genetics of mitochondrial DNA than its appearance became different. Usher-types syndrome and Kearns-Sayre are characterized to be hereditary like mitochondrial genes [29].

## SYNDROMIC RETINIS PIGMENTOSA

In case of syndromic RP ocular degeneration causes abnormalities in more than one or more organ system [30]. Most general forms of syndromic RP are Usher syndrome. The connection of retinitis pigmentosa and inherited sensorineural loss of hearing in that condition when other symptoms are not involved is recognized as usher syndrome. It is then identified as it has minimum two or may be three kind of usher syndrome, these form exist as Autosomal recessive form [31,32]. Usher syndrome type 1 has intense hereditary, bilateral neurosensory deafness and no understandable communication. Every affected person face the problems of vestibular nerve role in the caloric testing that causes the slow, non-progressive ataxia. Indication of usual RP are generally observed in delayed childhood to the start of puberty and then progress slowly, consequential in considerable loss of vision at the age of the mid-30's to mid of 40 years [30].

## BARDIET-BIEDL SYNDROME

BBS(Bardet Biedl syndrome) is less common form as compared to the usher syndrome which has the prevalence 1 in 150,000 [31]. Phenotype is feature that links the RP with fatness in the early age, mental retardation, post axial polydactyl renal abnormalities and, hypogenitalism all these causes the renal malfunction. In case of BBS minimum 11 mutated genes are observed [32,33], which shows triallelic digenic mode of inheritance [34].

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