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Hutchinson-Gilford Progeria Syndrome: A Comprehensive Review of Pathogenesis, Clinical features, Diagnosis and Management

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Abstract: Hutchinson–Gilford Progeria Syndrome (HGPS) is an extremely rare, fatal genetic disorder characterized by accelerated aging during early childhood. The condition is primarily caused by a de novo mutation in the LMNA gene (c.1824C>T), resulting in the production of progeria, an abnormal lamin A protein that disrupts nuclear architecture, genomic stability, and cellular function. Children with HGPS present with growth retardation, alopecia, lipodystrophy, osteoporosis, joint contractures, scleroderma-like skin changes, and characteristic facial features while maintaining normal cognitive development. Progressive cardiovascular disease, including atherosclerosis, myocardial infarction, heart failure, and stroke, remains the leading cause of mortality. Diagnosis is established through clinical evaluation and confirmation of LMNA mutations using molecular genetic techniques such as DNA sequencing and whole-exome sequencing. Current management is primarily supportive and focuses on symptom control and prevention of complications. The farnesyltransferase inhibitor lonafarnib is the only FDA-approved treatment and has demonstrated improved survival and cardiovascular outcomes. Emerging therapeutic approaches, including CRISPR/Cas9 gene editing, antisense oligonucleotides, RNA interference, mTOR inhibitors, and base-editing technologies, offer promising strategies for reducing progerin accumulation and correcting the underlying genetic defect. Continued research into targeted molecular therapies may improve survival, quality of life, and provide potential curative options for patients with HGPS.

Keywords: Hutchinson–Gilford Progeria Syndrome (HGPS), Progeria, LMNA gene, Progerin, Premature aging, Lamin A, Lonafarnib, Farnesyltransferase inhibitor, Gene therapy, CRISPR/Cas9, Cardiovascular complications, Rare genetic disorder.

Introduction

Progeroid syndrome, or PS for short, is a collection of uncommon and potentially fatal autosomal dominant genetic disorders that cause premature ageing.**(1)** Premature ageing disorders, also known as progeroid syndromes, are a diverse set of uncommon, severely deadly, and genetic illnesses that seem to mimic several signs of advanced physiological ageing very early in life.**(2)** A rare and deadly hereditary condition known as Hutchinson-Gilford progeria syndrome (HGPS) is brought on by a de novo point mutation in exon 11 of the LMNA gene (c.1824C > T; p.G608G). Progerin, a poisonous form of lamin A

with a 50-amino acid deletion close to its C terminus, is the outcome of this mutation. Progerin is constantly farnesylated as a result of this loss, making it incapable of undergoing normal lamin A processing. Disrupted gene expression, genomic instability, nuclear abnormalities, and altered redox balance are the outcomes of farnesylated progerin buildup in the nuclear lamina. **(3)** Only 400 children worldwide are thought to have this condition, which has an incidence of 1 in 20,000,000 births (Progeria Research Foundation). However, understanding the molecular pathways producing progerin toxicity is relevant for normal ageing, as progerin is thought to be produced to some extent in ageing normal individuals due to spontaneous activation of the cryptic splice site that is triggered in HGPS patients. It's interesting to note that progerin expression was absent from a study on centenarians. The functional link between progerin levels and lifespan and healthspan during normal human ageing should be the subject of further research, according to these findings. **(9)**

Symptoms

Alopecia, osteoporosis, atypical skin, loss of subcutaneous fat, joint contractures, and increasing atherosclerosis are some of the clinical characteristics that define this condition. For these individuals, the primary cause of death is complications from severe atherosclerosis, including heart failure, myocardial infarction, and cerebrovascular illness (stroke)**. (3, 19)** Loss of subcutaneous fat and muscle mass, bulging joints and reduced range of motion, and fingertip hypertrophy are some of the symptoms that affect the trunk and extremities. However, the majority of the impacted kids are lively and endearing, and there is no mental illness or intellectual disability. The majority of children over 12 weight about 15 kg, which is the same as that of healthy children aged 3 to 4. This growth disturbance is particularly noticeable in terms of weight.

They consequently gradually begin to resemble elderly people **(4)**. A unique face, large eyes, leg and scalp veins, a senile appearance, hair loss on the scalp, eyebrows, and eyelashes, stunted growth, and sclerodermatous alterations are some of the symptoms. Progerin, an aberrant form of the nuclear membrane protein Lamin A, accumulates when the LMNA gene is mutated. Although premature atherosclerotic cardiovascular disease is the primary cause of death for children, this accumulation can result in a variety of disease morphologies. The Progeria Research Foundation has been spearheading clinical trials and research in an attempt to discover a treatment and cure for this illness. **(1)**

Progeria case in India

According to the Progeria Research Foundation's most recent data, there are approximately 80 known documented occurrences of Progeria Syndrome globally, with 18 occurring in the US and 4 in India. In addition, it was anticipated that there were between 200 and 250 cases, of which about 60 were believed to originate from India. Four cases of Progeria have been discovered in India: Prachi from Patna, Nihal Bitla from Bhiwandi, five-year-old Aditya from Rajasthan, and Ali Hussain from Bihar. **(8)**

Diagnosis

Although apparent and prominent symptoms may appear later, the diagnosis can be made within the first six months of life. In the present review, the identification of progerin as a cause of HGPS is discussed, along with proof of its harmful effects when intracellularly produced and the advantages of blocking its expression. It also presents initiatives for HGPS clinical trials and treatment development. **(5)** Diagnostic requirements for HGPS include phenotype recognition and a progerin-producing mutation in the LMNA gene, either within exon 11 (conventional form) or near the exon 11 intronic boundary (atypical form). One LMNA gene is known to be the cause of HGPS, a completely penetrant autosomal dominant disorder. **(20)**

Despite the one known instance of somatic and gonadal mosaicism, HGPS is often caused by a de novo mutation. By sequencing the entire coding region and the splice junctions that link it, one can identify the common mutation of traditional HGPS as well as the mutations identifying atypical HGPS. Whole - exome sequencing (WES) of previously examined case reports showed synonymous heterozygous variance in the LMNA gene. One of these changes, C.1824 C>T (P. G608G), was spontaneous and synonymous and may alter the function of the gene. **(1)**

Management

Clinically, treating symptoms and preventing secondary consequences are key components of illness management. For example, congestive heart failure is treated with ant congestive treatment (Gordon, Brown, & Collins, 1993). Physical therapy, body bracing, or even reconstructive hip surgery can be used to address hip dislocation (Gordon *et al.,* 1993).

Those treatments are primarily palliative in nature, even though they may lessen disease symptoms or enhance patients' quality of life. Therefore, there is an urgent need to develop therapy options that can effectively treat the condition. The most recent advancements and efficacy of treatment approaches documented throughout the past ten years are methodically reviewed in this essay. **(6)**

The efforts can be broadly divided into gene therapy strategies that aim to use adenosine base editor techniques and CRISPR/Cas9 to correct the LMNA mutation, stimulate progerin degradation, inhibit the enzymes that post translationally modify progerin and prelamin A, and use antisense strategies that target abnormal splicing to decrease progerin mRNA expression.

Recent research has shown that ICMT is a possible enzymatic therapeutic target, however the majority of the latter efforts have concentrated on FTase. **(7)** A number of therapeutic approaches have been put forth to address the deficiencies in HGPS, such as:

(i) Directly "repairing" the transmutation;

(ii) Preventing the formation of progerin mRNA through pre-mRNA mutant splicing;

(iii) Reducing the dangerous amounts of isoprenylated and methylated progerin;

(iv) Producing progerin removal; and

(v) Reducing the detrimental accumulation of progerin **(12).**

Complication

Cardio Vascular Complication

Progerin buildup, which impacts both cardiac electrophysiology and vascular integrity, is the main cause of the serious cardiovascular problems associated with HGPS. Heart block, ventricular arrhythmias, bradyarrhythmias, and other cardiac electrophysiological abnormalities that are common in HGPS are often caused by myocardial fibrosis, which disrupts normal electrical transmission. Autonomic dysfunction makes these alterations worse, resulting in longer QT intervals and a higher risk of sudden cardiac death. **(1)** A weakening of the heart muscle and an inability to transmit impulses are the causes of cardiomyopathies and conduction abnormalities, which impair heart function. Based on tissue characteristics, cardiomyopathies are divided into three kinds. In hypertrophic cardiomyopathies (HCM), the left ventricle thickens and enlarges, resulting in stiff ventricles. Dilated cardiomyopathies (DCM) are caused by weak cardiac muscles that are unable to contract following diastole. Restrictive cardiomyopathies (RCM), in which the heart cannot relax its muscle after systole, result in an enlarged atrium**. (21, 25)**

Approximately 64% of patients with LMNA mutations had DCM with conduction abnormalities. Furthermore, a few studies found that uncontrolled cardiac dysrhythmias are linked to patients with dilated cardiomyopathy who are more likely to experience sudden cardiac death. There is evidence to support the theory that lamin mutations cause cardiac diseases, which impair heart function as people age **(10).**

Restrictive Dermopathy

Usually brought on by mutations in the ZMPSTE24 gene, this restricted dermatopathy, commonly referred to as "lethal hyperkeratosis-contracture syndrome," can also result from heterozygous de novo mutations in the LMNA gene. Premature birth results from foetal akinesia and polyhydramnios caused by intrauterine growth limitation. The look of affected persons is normal, with large nipples, erosions, scales, and thin, taut skin with hyperkeratosis. Their distinctive facial features include a short nose, low-set ears, a prominent inner corner of the eyes, and an O-shaped mouth. Contractures, dysplastic clavicles, and very

long bones are examples of bone abnormalities. The offspring pass away within the first week because of pulmonary hypoplasia caused by the foetal akinesia-deformation sequence. **(11)**

Mandibuloacraal Dysplasia

The underlying mutations in LMNA (MADA) or ZMPSTE24 (MADB), which both result in an accumulation of prelamin A and hence affect chromatin physiology due to inadequate lamin A, are used to categorise autosomal recessive mandibuloacral dysplasia (MAD). Local osteolysis (hypoplasia of the mandible, clavicle, fingers, and receding chin) and osteoporosis are characteristic, along with postnatal progeroid signs. The main difference between the two groups is that type A has fat deposits in the face and neck and partial lipodystrophy in the extremities, while MADB has more extensive lipodystrophy. **(11, 22)**

Nestor Guillermo Progeria Syndrome

Mutations in BANF1 (also known as "barrier to auto integration factor 1") result in the "Néstor–Guillermo progeria syndrome" (NGPS), which is characterised by a slower clinical course and a prolonged life expectancy. In addition to interacting with LMNA, BANF contributes to the nuclear lamina's construction and breakdown during mitosis. Only three examples have been documented thus far, and all of them had typical development until they were two years old. Nonetheless, there are striking similarities between the clinical appearance and the previously listed MAD disorders. Along with the previously noted progeroid signs, there is growth disruption and progressive lipoatrophy. Cardiovascular problems, which are typical in HGPS, were not seen despite the substantial skeletal alterations. **(11)**

Lonafarnib differentially affects elastic and muscular arteries

Elastic (e.g. the aorta) and muscular (e.g. branch mesenteric) arteries remodel differently in hypertension and natural aging. We thus quantified the biomechanical phenotype of the second-order branch mesenteric artery (MA) as a representative muscular artery, again comparing age-matched wild-type and progeria vessels from P42 to P168. Both structural (pressure-radius and axial force-stretch) and material stiffness were compromised by progeria despite no apparent histological signs of an excessive accumulation of proteoglycans or calcification. **(15)**

Pathophysiology

The majority of the de novo point mutations that cause HGPS, a rare autosomal dominant condition, occur in codon 608 of exon 11 on chromosome 1. The nuclear laminae, a complex molecular interface found within the inner nuclear membrane, are made up of three proteins known as lamin A (LA), lamin C (LC), and lamin 10. The LMNA gene encodes these proteins. It has now been established how important the lamina is for transcription, nuclear morphology, DNA replication, chromatin structure, and cell division. The most common type of LMNA mutation in HGPS is a C-T nucleotide change that creates a cryptic splice donor site at position 1824 without altering the encoded amino acids. This location causes mRNA to lack 150 nucleotides when it is active.

This mRNA is then translated into the "progerin" mutant protein, which has an internal deletion of 50 amino acids surrounding the C terminus. The integrity of the nuclear structure and function is impacted by LA, which is expressed by most differentiated cells. Progerin most likely affects the nuclear function of the cells that express LA in a dominant-negative manner. It is also thought to interfere with other vital functions like gene transcription, DNA replication, and cell division. A cysteine molecule with two aliphatic amino acids and a terminal "X" residue is known as a CAAX motif. The CAAX tetrapeptide motif is typically found in the C-terminus of prelamin A. With the aid of the enzyme farnesyltransferase (FTase), the tetrapeptide precisely instructs the cysteine to integrate a 15-carbon farnesyl isoprenoid lipid group.

With the aid of FTase signalling modification, the amino acids of the CAAX exactly specify the addition of an isoprenyl group with alanine, glutamine, methionine, or serine. Alternatively, a 20-carbon geranylgeranyl isoprenoid group will be added to leucine, as indicated by the geranylgeranyl transferase (GGTase) enzyme. CSIM is the CAAX motif for LA.

Prelamin farnesylation and the associated change in CAAX signalling increase contact with the nuclear membrane. After farnesylation, the three-terminal amino acids (AAX) are eliminated and the c-terminal cysteine group is methyl esterified. Remarkably, the mature protein loses the farnesylated cysteine and the additional 15 C-terminal amino acids due to a second cleavage that takes place inside the nucleus. Following this final cleavage step, the farnesyl anchor is removed, and the prelamin A is probably liberated from the nuclear membrane and integrated into the nuclear lamina. The endoprotease recognition region necessary for the last cleavage step is eliminated by the internal loss of amino acids 606–656, even though preprogerin can be farnesylated in HGPS.

The significance of this cleavage is demonstrated by the fact that transmutations in ZMPSTE24 cause a severe form of mandibuloacral dysostosis, one of several laminopathies that have clinical characteristics similar to HGPS. The ultimate cleavage of Lamin A depends on ZMPSTE24, the human equivalent of yeast STE24. Progerin retains its farnesyl group, which stabilises its connections with the inner nuclear membrane, since LMNA G608G stops the final cleavage. The nuclear envelope (NE) grows inward in the presence of progerin, increasing the surface area available to contain the excess progerin accumulation in HGPS. **(12)**

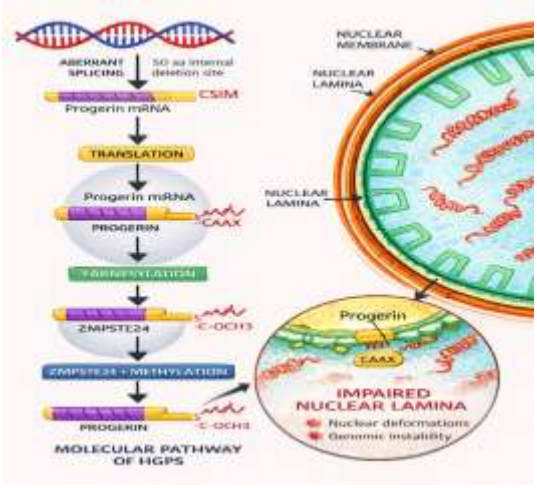

Fig 1: The Pathophysiological mechanisms of HGPS. (12)

Treatment

Two therapeutic approaches to the treatment of HGPS have been investigated in clinical trials. The first seeks to prevent prenylation and, therefore, incorporation of progerin into the nuclear membrane through the use of the farnesyltransferase inhibitor lonafarnib. Despite notable gastrointestinal side effects, prolonged treatment with lonafarnib resulted in decreased mortality rate of HGPS patients. The second approach, currently the topic of a clinical trial with everolimus, utilizes mammalian target of rapamycin (mTOR) inhibitors to facilitate lysosomal degradation of intracellular progerin by activation of autophagy. The broader effects of mTOR inhibition result in immunosuppression. Thus, both of these therapeutics include side effects that could exacerbate a condition of failure to thrive. These limitations of current approaches to HGPS highlight the persistent need for precisely targeted gene therapeutic techniques**. (16)**

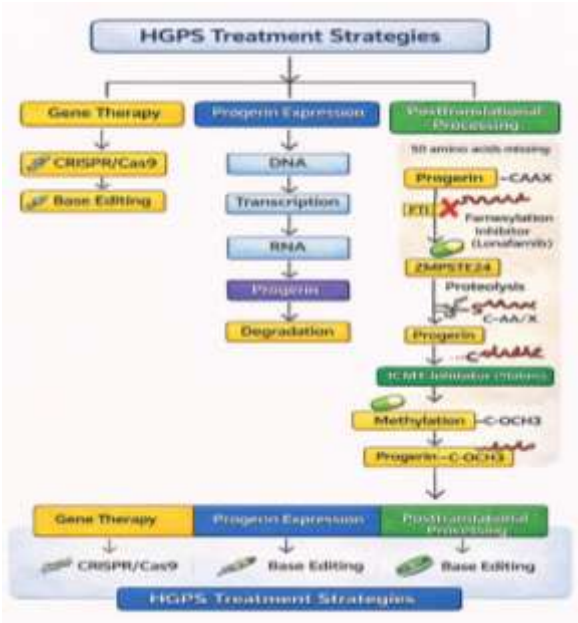

Fig 2: Overview of potential HGPS treatment strategies. (7)

Gene Therapy

Various strategies for HGPS gene therapy have already been designed. Some of them were based on gene editing approaches (CRISPR-Cas or adenine base editors) or on RNAi, whereas others demonstrate splice site blocking using antisense oligonucleotides (ASOs). One of the very first strategies was tested in patients' fibroblasts; it was based on lentiviral vectors and the overexpression of small hairpin RNA (shRNA) specifically targeting the unspliced progerin pre-mRNA or mature progerin. Ultimately, only one shRNA sequence, which recognizes the junction of progerin mRNA exons 11–12, was chosen. A decrease in the progerin level to 26% of that in nontransfected patients' fibroblasts and a nonsignificant reduction in the lamin A level were observed. Small interfering RNAs (siRNAs) have never been used to date in studies aimed at progerin silencing. However, they have several advantages over other tools used in gene therapy. **(17)**

Progerin levels can also be reduced by accelerating its degradation rate. Rapamycin—an inhibitor of mTOR signalling pathway—was found to reduce the characteristic nuclear shape abnormalities and delay senescence of HGPS cells in vitro. The effect was linked to increased progerin clearance. A clinical trial (NCT02579044) where everolimus, a rapamycin derivative, was given together with lonafarnib (i.e. the standard FTI therapy) was initiated to evaluate its efficiency in treating HGPS and other progeroid laminopathies. However, no results have yet been released from this trial. A potential limitation of this strategy is that rapamycin inhibits proliferation of human and mouse HGPS cells and worsens the senescence phenotype, at least in cultured cells. **(7)**

Post Translational Processing

Numerous investigations have shown that FTIs reduced the amount of Progerin and prevented its accumulation, supporting the idea that farnesyltransferase (FT) could be a therapeutic target for HGPS. They may have impeded the development of HGPS and processing-deficient progeroid laminopathies. **(5)** Nuclear blebbing or deformed nuclei, thickened nuclear lamina, and decreased mechanotransduction are the effects of permanent farnesylation of progerin, which binds the protein to the nuclear lamina. Studies on HGPS cells and mice show that progerin's farnesyl group plays a role in the phenotype of HGPS.

The farnesyltransferase inhibitor (FTI) lonafarnib decreases progerin intercalation into the nuclear envelope by preventing post-translational farnesylation of progerin. FTI therapy improves skeletal features, increases body weight and fat tissue, promotes cardiovascular phenotype, increases longevity, and improves nuclear morphology in HGPS cells in animal models. A clinical investigation found that

lonafarnib medication increased body weight, reduced cardiovascular stiffness, increased bone density, and reduced trial participant mortality.

As of right now, the only FDA-approved treatment for HGPS is lonafarnib. **(13)** The incidence of gastrointestinal tract (GIT) problems caused by lonafarnib may be decreased by taking 115 mg/m2 twice a day with breakfast and dinner.

The dosage may be increased to 150 mg/m2 twice daily after four months. For patients whose body surface area is less than 0.39 m2, there are no lonafarnib dose forms available. . Lonafarnib also improved cardiovascular stiffness, audiological conditions, and bone weight-bearing ability and fracture resistance; nonetheless, some people saw significant weight gain. Since the main cause of death for children with HGPS is heart disease, lonafarnib also resulted in a discernible improvement in heart health. Echo-dense common carotid arteries and a noticeably higher pulse wave velocity before treatment indicated that peripheral arterial stiffness had improved following therapy.

There was also an improvement in low-level sensorineural hearing. Numerous clinical studies have shown that FTIs can cause prelamin A to build up and are generally well tolerated, with side effects usually limited to the gastrointestinal system**. (1)**

A critical posttranslational processing step for protein intercalation into the inner nuclear membrane is progerin farnesylation, which is inhibited by farnesyltransferase inhibitors (FTIs), which are tiny compounds that reversibly bind to the farnesyltransferase CAAX binding site. Progerin-containing cells regain their normal shape and function after receiving FTI therapy in vitro. Furthermore, FTIs ameliorate cardiovascular disease and increase lifespan in the mouse transgenic model of HGPS. A phase II study of the FTI lonafarnib's main goals were either a 50% increase in weight gain over the pretherapy-predicted annual rate or a shift from weight loss to statistically meaningful weight gain while on treatment. In this study, we looked at neurologic characteristics both prior to and during the start of oral lonafarnib treatment. **(14,23)**

Triple Therapy

Blocking progerin, geranylgeranylation, and farnesylation of prelamin A can avoid HGPS symptoms, according to a comprehensive clinical investigation including 45 HGPS patients receiving a combination of lonafarnib, pravastatin, and zoledronate medication. Additionally, pravastatin slows the growth of atherosclerosis and zoledronate inhibits osteoporosis. The carotid artery echodensity and/or the per-patient weight increase improved, and almost 71.0% of patients satisfied the predefined outcome criteria. In HGPS, lonafarnib monotherapy and triple therapy showed an average increase in survival of 1.6 years**. (1)**

Dosage and Administration.

After at least four months, lonafarnib (Merck & Co., Inc., Whitehouse Station, NJ) was started at 115 mg/m2 and increased to 150 mg/m2 if tolerated. For 24 to 29 months, participants were given oral lonafarnib twice a day in the form of capsules or liquid solution. **(14)**

Treatment with the farnesyltransferase inhibitor lonafarnib significantly reduced mortality in patients with Hutchinson–Gilford Progeria Syndrome (HGPS). In the ProLon1 trial, 27 genetically confirmed LMNA patients received oral lonafarnib (115 mg/m² twice daily, increased to 150 mg/m²), showing lower mortality (3.7%) compared to matched untreated controls (33.3%). Similarly, combined analysis of ProLon1 and ProLon2 (63 patients) showed reduced deaths in treated patients (6.3%) versus untreated controls (27.0%). Most deaths (79%) were attributed to heart failure, highlighting cardiovascular complications as the primary cause of mortality in HGPS. **(24)**

Conclusion

Targeting the post-translational farnesylation of progerin using farnesyltransferase inhibitors (FTIs) such as lonafarnib has demonstrated clinical benefits, including improvements in vascular stiffness, weight gain, and overall survival. This underscores the crucial role of aberrant post-translational modification in the disease mechanism and highlights a viable therapeutic target for intervention.

Increased survival:

- Median survival increased by 2.5 years compared to untreated historical controls (JAMA, 2018).

Improved vascular health:

- Decreased arterial pulse wave velocity (a measure of arterial stiffness).
- Reduced carotid artery echodensity (suggesting healthier arterial walls).

Weight gain and growth:

- Significant weight gain in many patients, reversing typical growth failure in HGPS.

Bone health and mobility:

- Reduced skeletal rigidity and improved joint mobility.

Hearing improvements:

- Partial recovery from sensorineural hearing loss.

Neurological improvements:

- Fewer headaches**,** transient ischemic attacks (TIAs)**,** and strokes**.**

Lonafarnib significantly reduces mortality in children with HGPS compared to historically matched untreated groups, with hazard ratios ranging from 0.12 to 0.23—indicating approximately an 80–88% reduction in death risk during the studied time frames. Survival extension ranges from 1.6 to 2.5 years, depending on the duration of follow-up and study cohort.

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