Bibliography

Below is a listing of some recommended reading on Progeria. The list highlights many of the points made within the body of this handbook. It is by no means exhaustive. For additional reading, we recommend you go to PUBMED and search Progeria, lamin, or laminopathy. Some of the articles that your search finds will be free for downloading.

Websites

www.progeriaresearch.org/patient_care.html
The Progeria Handbook: A Guide for Families & Health Care Providers of Children with Progeria
Clinical guidelines by system, psychosocial strategies, basic science and genetics

www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=hgps
GeneReviews - A general clinical and genetics and basic science review

www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=176670
On Mendelian Inheritance in Man (OMIM) – Detailed high level genetics and landmark articles

www.clinicaltrials.gov/ct2/results?term=progeria
Clinical Trials Information

www.progeriaresearch.org/patient_registry.html
PRF International Patient Registry

www.progeriaresearch.org/diagnostic_testing.html
PRF Diagnostic Testing Program

www.progeriaresearch.org/medical_database.html
PRF Medical & Research Database

www.progeriaresearch.org/cell_tissue_bank.html
PRF Cell & Tissue Bank

Reviews and book chapters

Gordon, LB. The Premature Aging Syndrome Hutchinson-Gilford Progeria Syndrome: Insights Into Normal Aging. In H. M. Fillit, K. Rockwood, K. Woodhouse (Eds.) Brocklehurst's Textbook of Geriatric Medicine and Gerontology (7th ed.). W.B. Saunders, Elsevier 2010;66-72.

Rodriguez S, Eriksson M. Evidence for the Involvement of Lamins in Aging. Curr Aging Sci 2010.

Capell BC, Tlougan BE, Orlow SJ. From the Rarest to the Most Common: Insights from Progeroid Syndromes into Skin Cancer and Aging. J Invest Dermatol 2009.

Gordon, LB, Brown, WT, Rothman, FG. LMNA and the Hutchinson-Gilford Progeria Syndrome and Associated Laminopathies. In C. J. Epstein, R. P. Erickson, A. Wynshaw-Boris (Eds.) Inborn Errors of Development: The molecular basis of clinical disorders of morphogenesis (2nd ed.). New York, NY: Oxford University Press 2008;139:1219-1229.

Kieran, MW., Gordon, LB, Kleinman, M. New Approaches To Progeria. State-Of-The-Art Review Article. Pediatrics Oct2007;120(4):834-41.

Capell BC, Collins FS, Nabel EG. Mechanisms of cardiovascular disease in accelerated aging syndromes. Circ Res 2007;101(1):13-26.

Capell BC, Collins FS. Human laminopathies: nuclei gone genetically awry. Nat Rev Genet 2006;7(12):940-52.

DeBusk FL. The Hutchinson-Gilford progeria syndrome. Report of 4 cases and review of the literature. J Pediatr 1972;80(4):697-724

Primary Research Articles

> Global Clinical Studies on Progeria:

Hennekam RC. Hutchinson-Gilford Progeria syndrome: review of the phenotype. Am J Med Genet A 2006;140(23):2603-24.

Merideth MA, Gordon LB, Clauss S, Sachdev V, Smith AC, Perry MB, et al. Phenotype and course of Hutchinson-Gilford Progeria syndrome. N Engl J Med 2008;358(6):592-604.

> Subspecialty Studies on Progeria:

Anesthesia: Liessmann CD. Anaesthesia in a child with Hutchinson-Gildford progeria. Paediatr Anaesth 2001;11(5):611-4.

Dental: Domingo DL, Trujillo MI, Council SE, Merideth MA, Gordon LB, Wu T, et al. Hutchinson-Gilford Progeria syndrome: oral and craniofacial phenotypes. Oral Dis 2009;15(3):187-95.

Growth and Bones: Gordon LB, McCarten KM, Giobbie-Hurder A, Machan JT, Campbell SE, Berns SD, et al. Disease progression in Hutchinson-Gilford Progeria syndrome: impact on growth and development. Pediatrics 2007;120(4):824-33.

Dermatology: Gillar PJ, Kaye CI, McCourt JW. Progressive early dermatologic changes in Hutchinson-Gilford progeria syndrome. Pediatr Dermatol 1991;8(3):199-206.

Growth Hormone: Sadeghi-Nejad A, Demmer L. Growth hormone therapy in progeria. J Pediatr Endocrinol Metab 2007;20(5):633-7.

> Aging and Progeria:

McClintock D, Ratner D, Lokuge M, Owens DM, Gordon LB, Collins FS, et al. The mutant form of lamin A that causes Hutchinson-Gilford Progeria is a biomarker of cellular aging in human skin. PLoS One 2007;2(12):e1269.

Scaffidi P, Gordon L, Misteli T. The cell nucleus and aging: tantalizing clues and hopeful promises. PLoS Biol 2005;3(11):e395.

> Genetics – Discovery:

De Sandre-Giovannoli A, Bernard R, Cau P, Navarro C, Amiel J, Boccaccio I, et al. Lamin a truncation in Hutchinson-Gilford progeria. Science 2003;300(5628):2055.

Eriksson M, Brown WT, Gordon LB, Glynn MW, Singer J, Scott L, et al. Recurrent de novo point mutations in lamin A cause Hutchinson-Gilford Progeria syndrome. Nature 2003;423(6937):293-8.

> Cell Shape:

Goldman RD, Shumaker DK, Erdos MR, Eriksson M, Goldman AE, Gordon LB, et al. Accumulation of mutant lamin A causes progressive changes in nuclear architecture in Hutchinson-Gilford progeria syndrome. Proc Natl Acad Sci USA 2004;101(24):8963-8.

> Treatments in Cells:

Yang SH, Bergo MO, Toth JI, Qiao X, Hu Y, Sandoval S, et al. Blocking protein farnesyltransferase improves nuclear blebbing in mouse fibroblasts with a targeted Hutchinson-Gilford progeria syndrome mutation. Proc Natl Acad Sci USA 2005;102(29):10291-6.

Glynn MW, Glover TW. Incomplete processing of mutant lamin A in Hutchinson-Gilford progeria leads to nuclear abnormalities, which are reversed by farnesyltransferase inhibition. Hum Mol Genet 2005;14(20):2959-69.

Capell BC, Erdos MR, Madigan JP, Fiordalisi JJ, Varga R, Conneely KN, et al. Inhibiting farnesylation of progerin prevents the characteristic nuclear blebbing of Hutchinson-Gilford progeria syndrome. Proc Natl Acad Sci USA 2005;102(36):12879-84.

Mallampalli MP, Huyer G, Bendale P, Gelb MH, Michaelis S. Inhibiting farnesylation reverses the nuclear morphology defect in a HeLa cell model for Hutchinson-Gilford progeria syndrome. Proc Natl Acad Sci USA 2005;102(40):14416-21.

> Treatments in Mice:

Yang SH, Meta M, Qiao X, Frost D, Bauch J, Coffinier C, et al. A farnesyltransferase inhibitor improves disease phenotypes in mice with a Hutchinson-Gilford progeria syndrome mutation. J Clin Invest 2006;116(8):2115-21.

Capell BC, Olive M, Erdos MR, Cao K, Faddah DA, Tavarez UL, et al. A farnesyltransferase inhibitor prevents both the onset and late progression of cardiovascular disease in a progeria mouse model. Proc Natl Acad Sci USA 2008;105(41):15902-7.

Varela I, Pereira S, Ugalde AP, Navarro CL, Suarez MF, Cau P, et al. Combined treatment with statins and aminobisphosphonates extends longevity in a mouse model of human premature aging. Nat Med 2008;14(7):767-72.