GENETICS AND GENODERMATOSES

EARLY DIAGNOSIS OF HUTCHINSON-GILFORD PROGERIA SYNDROME WITH LMNA MUTATION

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Background: Hutchinson-Gilford progeria syndrom(HGPS) is a rare disorder with premature aging of the skin, bones, heart, and blood vessels. HGPS is a genetic disease typically caused by mutations in LMNA. Expression of the mutant product is progerin, its accumulation elicits nuclear morphological abnormalities, misregulated gene expression, defects in DNA repair, telomere shortening and genomic instability, lead to limit of cellular proliferative capacity. We describe a 3-month-old girl with HGPS.

Observation: The girl appeared normal at birth with a full-term delivery, but she soon presented with sclerodermoid appearance within ten days. At three-month old, her skin of the trunk, hip, vulva and legs appeared swollen and sclerodermoid, her knee and ankle joints contracted and her hair was sparse. The girl's manifestations suggest the possible diagnosis of HGPS, while it should be identificated with generalized morphea, neonatal scleredema and skin stiff syndrome. LMNA genetic analysis of her peripheral blood DNA revealed typical heterozygous mutation (c.1824C>T, p. G608G), while her parents were normal. In one year's follow-up, she failed to thrive. Her swollen skin gradually became softened, but characteristic manifestation of progeria appeared such as alopecia, frontal bossing, prominent scalp veins, delayed closure of anterior fontanelle, small ear lobes, sclerodermoid skin, lipodystrophy, short stature and low weight. Her intelligence was normal. Complete blood count, urine test, lipids, serum electrolytes, liver and renal function, myocardial enzymes were normal. Serum biotin level was lower than normal. We provided oral biotin, calcium, vitamin D for her. Long-term follow-up is needed to observe cardiovascular and skeletal abnormalities.

Key message: We should be aware of the early cutaneous manifestations of HGPS. Genetic analysis could help us expedite diagnosis, minimize testing and long term followed-up, before the appearance of progeria became more apparent. We try to improve the quality of patient's life and extend their limited life.