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A case report: Ovarian new growth in progeria

Cherise Andrea E Llaneta

Ateneo de Manila University School of Medicine and Public Health, Philippines

The Hutchinson-Gilford Progeria Syndrome (HGPS) is a rare genetic disease that involves single-base gene mutation in the LMNA gene, which results in the production of a dysfunctional and mutant lamin A protein called Progerin. Progerin is found in increased concentration in normal older individuals hence, patients with increased amount of this protein present with phenotypic signs of aging. Aging phenotypes associated in the normal aging process and not present in HGPS include predisposition to forming masses, cancer, cataract, increased abdominal fat, and neurodegeneration. Progeria is a rare genetic condition with an estimated incidence of 1 in 4 million live births and with a prevalence of 1 in 20 million living individuals. This paper presents a rare case of an adolescent female patient with Progeria who manifested with an ovarian mass.

Based on current review of literature, there is no established predisposition and association between abdominal masses and Progeria patients, specifically ovarian masses in adolescent females. Research has also shown that Progeria patients are resistant to developing tumors or cancer due to the actions of BRD4 gene which inhibits tumorigenic potential of transformed cells. Furthermore, the mitochondrial dysfunction in HGPS leads to decreased ability to be dysplastic or anaplastic thereby making HGSP patients less likely to develop tumors and have cancer. The exact mechanism by which this patient became predisposed to developing an ovarian mass is still unknown. Further studies to establish the correlation between Hutchinson-Gilford progeria syndrome (HGPS) and abdominal masses, specifically masses in the reproductive system, has yet to be undertaken.

Through this case report, it can be suggested that routine abdominal ultrasound or abdominal CT scan can be done to screen for the presence of masses in HGPS patients, especially in adolescent females.

Biography

Cherise Andrea Llaneta demonstrated an early interest in science and medicine. She pursued this passion by obtaining her Bachelor's degree in BS Biology from Ateneo de Manila University. She continued her academic journey by earning a Doctor of Medicine (MD) from Ateneo School of Medicine and Public Health where she graduated with top honors as a merit scholar. Recognizing the importance of public health in her country, she pursued a Master's degree in Public Health and Business Administration as well. With a passion for advancing healthcare through cutting-edge research and a commitment to patient care, Cherise Llaneta has become a leading figure in the medical community.