New study aims to define the progeria phenome

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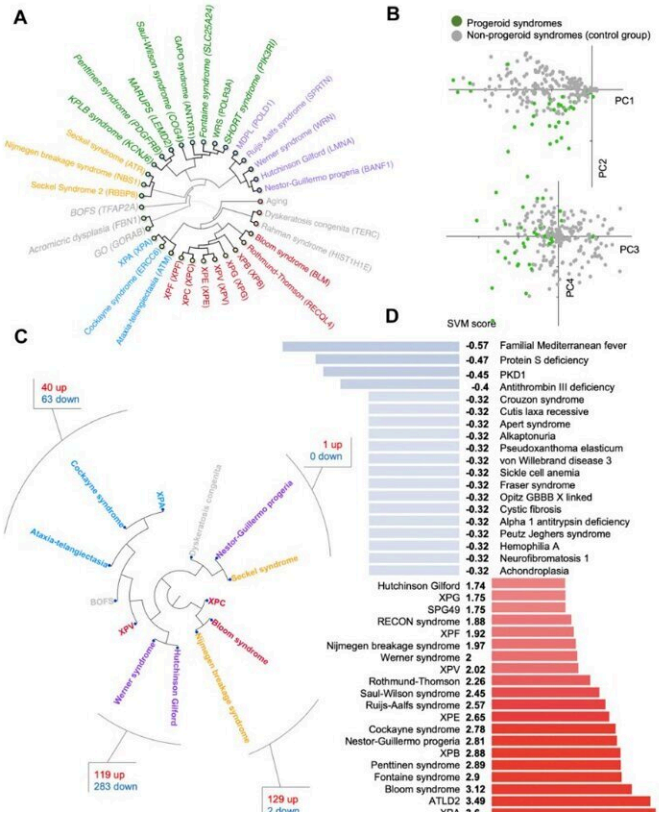

Progeroid syndrome overview. Credit: 2024 Worm et al.

Progeroid disorders are a heterogenous group of rare and complex hereditary syndromes presenting with pleiotropic phenotypes associated with normal aging. Due to the large variation in clinical presentation the

diseases pose a diagnostic challenge for clinicians which consequently restricts medical research.

In a new study, researcher Cecilie Worm, and others from the University of Copenhagen and Insilico Medicine aimed to accommodate this challenge by compiling a list of known progeroid syndromes and calculating the mean prevalence of their associated phenotypes, defining what they term the "progeria phenome." The work is published in the journal *Aging*.

"In this study, we have utilized phenome explorations to define the phenotypes associated with progerias and to develop tools to diagnose patients and identify new progeroid syndromes," the authors write.

The data were used to train a support vector machine that is able to classify progerias based on phenotypes. Furthermore, this allowed the researchers to investigate the correlation of progeroid syndromes and syndromes with various pathogenesis using hierarchical clustering algorithms and disease networks.

They detected that ataxia-telangiectasia like disorder 2, spastic paraplegia 49 and Meier-Gorlin syndrome display strong association to progeroid syndromes, thereby implying that the syndromes are previously unrecognized progerias.

"In conclusion, our study has provided tools to evaluate the likelihood of a syndrome or patient being progeroid. This is a considerable step forward in our understanding of what constitutes a premature aging disorder and how to diagnose them," say the researchers.

More information: Cecilie Worm et al, Defining the progeria phenome, *Aging* (2024). DOI: 10.18632/aging.205537