ABSTRACT FOR THE MARFAN FOUNDATION SYMPOSIUM

THE 101 GENOMES MARFAN PROJECT OF THE 101 GENOMES FOUNDATION

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OBJECTIVES: The 101 Genome Foundation (F101G) aims to create a **bioinformatics tool** containing the **genomic and phenotypic cross data** of patients with **rare diseases** (like Heritable Thoracic Aortic Disease (HTAD)). This tool, accessible to scientists through **a secure computer platform**, will allow researchers to reach a better comprehension of variable phenotypic expressivity in rare diseases. Moreover, it provides the opportunity to identify the existence of possible modifier genes that protect against (or aggravate) the major damage caused by the primary mutation in rare diseases. Identification of possible **protective modifier genes** could allow the development of treatments that replicate their protective effects in patients whose genes are not activated in the same way.

METHOD: The 101 Marfan Genome Project (P101GM) is the pilot project of the F101G and is dedicated to **Marfan syndrome (MFS)**. It is built on an **expandable starting cohort of 101 patients** with MFS. The P101GM will initially focus on **cardiovascular manifestations**, but is extendable in a later stage to other MFS related manifestations. Individuals harboring the same recurrent *FBN1* mutations but with variation in their cardiovascular phenotype are chosen for the composition of the initial cohort, after which the patients at the extreme ends of the phenotypical spectrum will be selected for further analysis. **WGS** data will be generated from the selected participants and stored in a secure computer platform.

CONCLUSION: The platform will allow researchers to better understand the clinical variability in MFS and identifying possible protective modifier genes that prevent cardiovascular manifestations caused by MFS.