P-078 - SIMPLE TOOLS FOR FABRY DISEASE PHENOTYPE IDENTIFICATION.

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INTRODUCTION: Fabry disease (FD) is an X-linked lysosomal storage disorder caused by mutations in the GLA gene. Two major phenotypes have been described: the type I “classic” and the type II “late-onset”. AIM: to describe simple tools for phenotype identification in clinical practice. METHODS: Review of medical records of patients diagnosed with FD during 16 years of follow up and comprehensive literature review. RESULTS: symptoms and signs description and age of onset is the first practical tool that clinicians may use for characterization. Late onset variants use to show cardiac or renal involvement after the third decade without the typical features described in children. Residual enzyme activity may correlates with the clinical phenotype, where less than 1% is related to classic phenotype. Mutation analysis using different databases and bioinformatic predictive tools also add other clue. Currently, plasmatic LysoGb3 measurement could be the second most important tool to define the phenotype and also may confirm the FD diagnosis. Lastly, endothelial involvement in tissue biopsy is only present in classic phenotype of FD. Description of intralysosomal inclusions in differentiated cells (podocytes, cardiomycites, etc) does not allow phenotype characterization. CONCLUSION: Phenotype identification in FD is mandatory because patient management, treatment prescription and clinical follow up are different in each case.