Retinal dystrophies are a group of rare diseases that damage the retina, the light-sensitive layer in the back of your eye. The retina sends signals to your brain so that you can see. Inherited means the condition runs in families. Often these diseases damage rods and cones in the retina.

SSBP1 is essential for mitochondrial DNA replication and maintenance, with defects leading to a spectrum of disease that includes optic atrophy and/or retinal dystrophy, occurring with or without extraocular features. A study of Jurkute et al. provided evidence of intrafamilial variability and confirms the existence of an autosomal recessive inheritance in SSBP1-disease consequent upon a previously unreported genotype [1)].

McArdle disease is caused by recessive mutations in PYGM gene. The condition is considered to cause a “pure” muscle phenotype with symptoms including exercise intolerance, inability to perform isometric activities, contracture, and acute rhabdomyolysis leading to acute renal failure. This is a retrospective observational study aiming to describe phenotypic and genotypic features of a large cohort of patients with McArdle disease between 2011 and 2019. Data relating to genotype and phenotype, including frequency of rhabdomyolysis, fixed muscle weakness, gout and comorbidities, inclusive of retinal disease (pattern retinal dystrophy) and thyroid disease, were collected. Data from 197 patients are presented. Seven previously unpublished PYGM mutations are described. Exercise intolerance (100%) and episodic rhabdomyolysis (75.6%) were the most common symptoms. Fixed muscle weakness was present in 82 (41.6%) subjects. Unexpectedly, ptosis was observed in 28 patients (14.2%). Hyperuricaemia was a common finding present in 88 subjects (44.7%), complicated by gout in 25% of cases. Thyroid dysfunction was described in 30 subjects (15.2%), and in 3 cases, papillary thyroid cancer was observed. Pattern retinal dystrophy was detected in 15 out of the 41 subjects that underwent an ophthalmic assessment (36.6%). In addition to fixed muscle weakness, ptosis was a relatively common finding. Surprisingly, dysfunction of thyroid and retinal abnormalities were relatively frequent comorbidities. Further studies are needed to better clarify this association, although our finding may have important implication for patient management [2)].

1)
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2)
Pizzamiglio C, Mahroo OA, Khan KN, Patasin M, Quinlivan R. Phenotype and genotype of 197 British patients with McArdle disease: An observational single-centre study. J Inherit Metab Dis. 2021 Nov;44(6):1409-1418. doi: 10.1002/jimd.12438. Epub 2021 Sep 22. PMID: 34534370.