

Retinitis pigmentosa

Retinitis pigmentosa - RP is a set of hereditary disorders characterized by a progressive loss of peripheral vision , which often leads to an incurable loss of central vision - blindness. vidění, které často vede k neléčitelné ztrátě centrálního visu – slepotě.

RP is known to include many types of retinal dystrofií and retinal pigment dystrophies due to molecular defects in more than 100 genes. At the same time, the label retinitis is somewhat misleading, because typical signs of inflammation do not primarily occur in the disease, it is a disorder of genes (and therefore also of their proteins), therefore even targeted therapy should be aimed at the molecular genetic level.

The disease most often occurs in isolation, but it can also be in association with another disease, the most common of which is deafness (up to 30% of patients), many people affected in this way have a diagnosis of Usher syndrome.