Supplementary Figure Legend. mTOR-related human syndromes

A series of germline mutations in genes involved in mTOR signaling pathways lead to different syndromes characterized by mTORC1 hyperactivity. With the exception of the syndrome caused by p14 mutation, these syndromes are caused by alterations in tumor suppressor genes, and consequently, are characterized by the spontaneous development of different tumors and other clinical manifestations. Images: gigantism of fingers in Proteus syndrome [1]; neurofibromas and skin pigmentation in neurofibromatosis [2]; intestinal tumor in LKB1 syndrome [3]; renal tumor in tuberous sclerosis [4]; phaeochromocytoma and pancreatic endocrine tumor in VHL [5]; short stature and hypopigmentation in p14-associated syndrome [6].

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Triglyceride: A lipid formed by the esterification fatty acids with glycerol and the most abundant form of lipid storage.

GEF: Acronym for Guanine nucleotide exchange factor, a protein that promotes GTP loading of G-proteins, resulting in their activation.

GAP: Acronym for GTPase activating protein, which promotes hydrolysis of GTP to GDP by G-proteins, resulting in their inactivation.

Macrolides: Naturally-occurring drugs, generally antibiotics, composed of a large lactone carbon ring.

Autophagosome: A transient membrane vesicle that engulfs and digests cellular components.

Gluconeogenesis: The chain of enzymatic reactions, mainly occurring in the liver, which leads to de novo production of glucose from more simple carbon precursors and ATP.

Supplementary Figure