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BBS5 Antibody Blocking Peptide

Catalog Number: bs-9438P

Activity: Not tested

Purification: HPLC

Storage: Shipped at 4°C. Stored at -20°C for one year. Avoid repeated freeze/thaw cycles.

Background: Bardet-Biedl syndrome (BBS) is a pleiotropic genetic disorder characterized by obesity,

photoreceptor degeneration, polydactyly, hypogenitalism, renal abnormalities, and developmental delay. Other associated clinical findings in BBS patients include diabetes, hypertension and congenital heart defects. BBS is a heterogeneous disorder; BBS genes map to eight genetic loci and encode eight proteins, BBS1-BBS8. Five BBS genes encode basal body or cilia proteins, suggesting that BBS is a ciliary dysfunction disorder. BBS5 localizes to ciliary basal bodies and is a member of the basal body/flagellar proteome. It plays a role in flagellar and basal body assembly and function. A mutation or loss of BBS5

may be correlated with photoreceptor degeneration