

**iBioSeminars: Part 3**  
Protein secretion and human disease

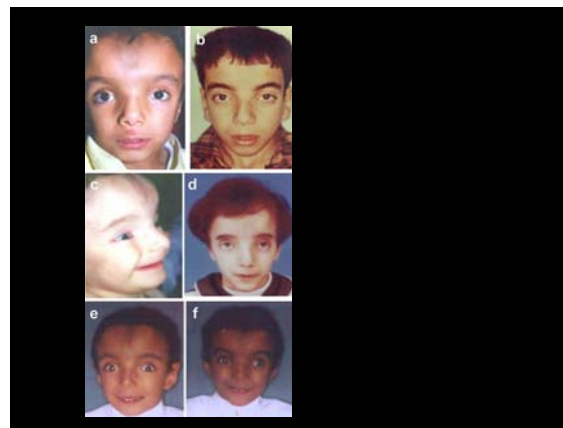
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**Mutations in a Sar1 GTPase of COPII vesicles are associated with lipid absorption disorders**

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Dietary fat is an important source of nutrition. Here we identify eight mutations in *SARA2* that are associated with three severe disorders of fat malabsorption. The Sar1 family of proteins initiates the intracellular transport of proteins in COPII (coat protein)-coated vesicles. Our data suggest that chylomicrons, which vastly exceed the size of typical COPII vesicles, are selectively recruited by the COPII machinery for transport through the secretory pathways of the cell.



**CLSD mutation: Alignment with yeast sequence and structure**

SEC23A	TGGYVMVGDSFNTSL <b>F</b> KQTFQRVFTKDMHGQFKMGF
SEC23B	TGGYVMVGDSFNTSL <b>F</b> KQTFQRIFTKDFNGDFRMAF
Sec23p	TGGVLLLTDAFSTAI <b>F</b> KQSYLRLFAKDEEGYLKMAF

