

ALSTROM SYNDROME

Marc E. Tischler, PhD;
University of Arizona

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- defective ALMS1 gene produces protein of unknown specific function
- protein plays diverse roles including:
 - hearing
 - vision {dystrophy of rod and cone cells in the retina}
 - regulating body weight {diminished satiety factor(s)}
 - heart function
 - kidney function
 - lung function
 - liver function
 - secretion of insulin by the pancreas
 - functioning of cilia that are involved in cell movement and chemical signaling
- protein located in centrosomes whose functions include:
 - division of cells
 - assembly of microtubules whose functions include:
 - transport of materials in cells (e.g., insulin movement to cell surface in pancreas)
 - maintenance of cell shape

