



Small G-protein & mentioned disease summary

❖ Summary of Diseases Mentioned in lecture (7,8,9):

➤ Muscular Dystrophy (related to actin filaments):

- *Duchenne Muscular Dystrophy (DMD)*: Caused by a **lack of functional dystrophin**, leading to progressive muscle degeneration,.
 - This DNA mutation is called “frameshift mutation”.
- *Becker Muscular Dystrophy (BMD)*: A milder form, caused by **partially functional** dystrophin (**dystrophin is shorter** than normal).

➤ Alzheimer’s Disease (linked to microtubules):

- Related to **tau** protein in microtubules (it’s a regulatory protein-MAP), which aggregates in neurons, contributing to cell death and dementia.
- Mutants in certain **kinesin proteins** can reduce the ability of neurons to move essential organelles leading to neurodegeneration such as Alzheimer’s Disease

➤ Amyotrophic Lateral Sclerosis (ALS), also known as Lou Gehrig’s disease, (connected to kinesin(microtubules) & neurofilaments (intermediate filaments)):

- caused by different factors like mutations in **lamin A (the IFs nuclear skeleton protein) OR** because of abnormalities in **microtubule motor protein (kinesin)**. ALSO, it can be caused by defective neurofilaments.

➤ Epidermolysis Bullosa Simplex (related to intermediate filaments):

- Caused by **keratin gene mutations**, resulting in fragile skin that blisters easily under mechanical stress.

➤ Charcot-Marie-Tooth Disease:

- Linked to mutations in **lamin A** (an intermediate filament) and **kinesin** (a microtubule motor protein), causing peripheral neuropathies.

❖ **Summary of Small G-protein mentioned in all lectures:**

<i>Small G Protein</i>	Function	Location
<i>Rab</i>	Determines vesicle membrane targets and ensures correct vesicular fusion	Vesicles (attached to vesicular membranes)
<i>Ran</i>	Regulates nuclear import/export via importin and exportin proteins	Nucleus and cytoplasm
<i>Ras</i>	Controls cell proliferation and differentiation through signaling pathways	Plasma membrane (at signaling sites)
<i>Rac</i>	Facilitates lamellipodia formation, aiding in cell movement and motility	Plasma membrane (leading edge of cells)
<i>Rho</i>	Promotes stress fiber formation and cell contraction	Plasma membrane (around actin filaments)
<i>Cdc42</i>	Initiates filopodia formation, aiding in cell directionality	Plasma membrane (cell periphery)
<i>Rab27A</i>	Involved in melanosome transport, as seen in Griscelli syndrome	Vesicles (specific to melanocyte function)