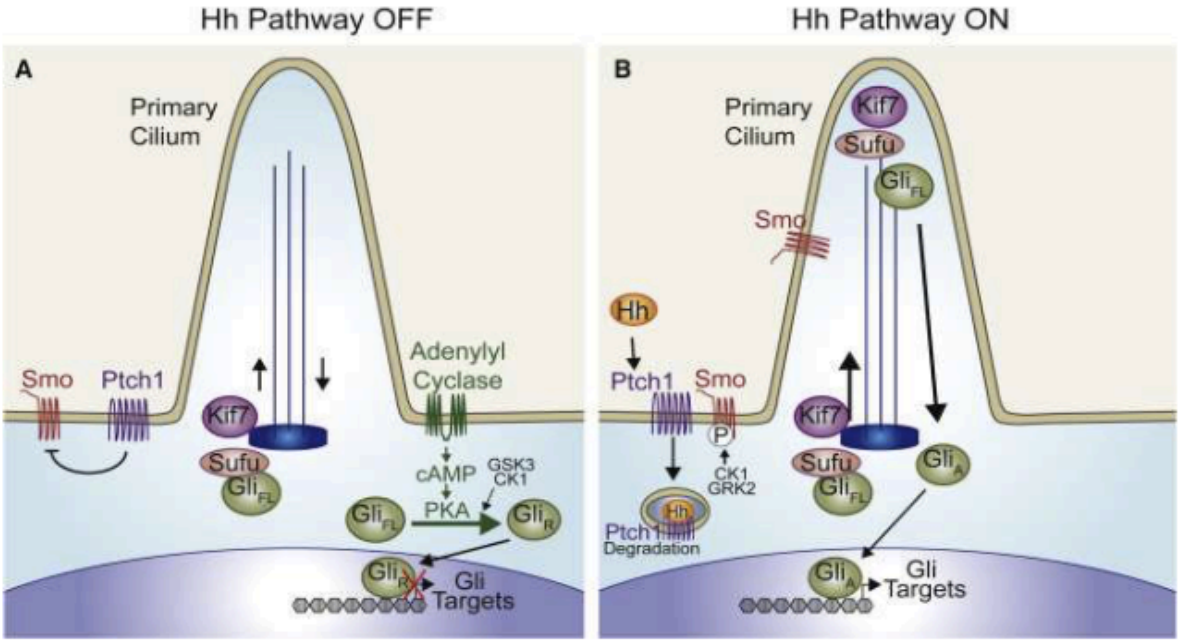

Signalling components moved by anterograde and retrograde transport within cilium

6. If you have a defect in the cilium or in the transport system, this will result in a defect in the hedgehog signalling pathway.
7. In your body, you have two types of cilium:
    - **Motile cilia –** found in the oviduct for example, where they move back and forth through the organs. They are characterised by the 9+2 axoneme.
    - **Primary cilium –** found in almost every organ in the body and one cilium per cell. The organelle that is housing a part of the hedgehog signalling network can be lost due to the defect in the pathway itself. These cilia are characterised by 9+0 axonemes.
8. As a result, we have a class of diseases classed as **ciliopathies,** such as polycystic kidney disease and retinitis pigmentosa (defect in the rods in the eyes), which is an example of an organ specific ciliopathy.
9. You can also have more widespread defects such as craniofacial abnormalities and polydactyly due to a loss or gain of hedgehog signalling.
10. Kartagener Syndrome is a result of motile cilium defects, which are lung or fertility defects.

Notch Signalling & Disease:

1. The notch pathway is like the Wnt & Shh pathway & its dysregulation is linked to many birth defects such as Alagille syndrome, where there are liver defects, CADASIL (cerebral autosomal dominant arteriopathy and subcortical infants and leukoencephalopathy), where there are defects in smooth muscle development.