

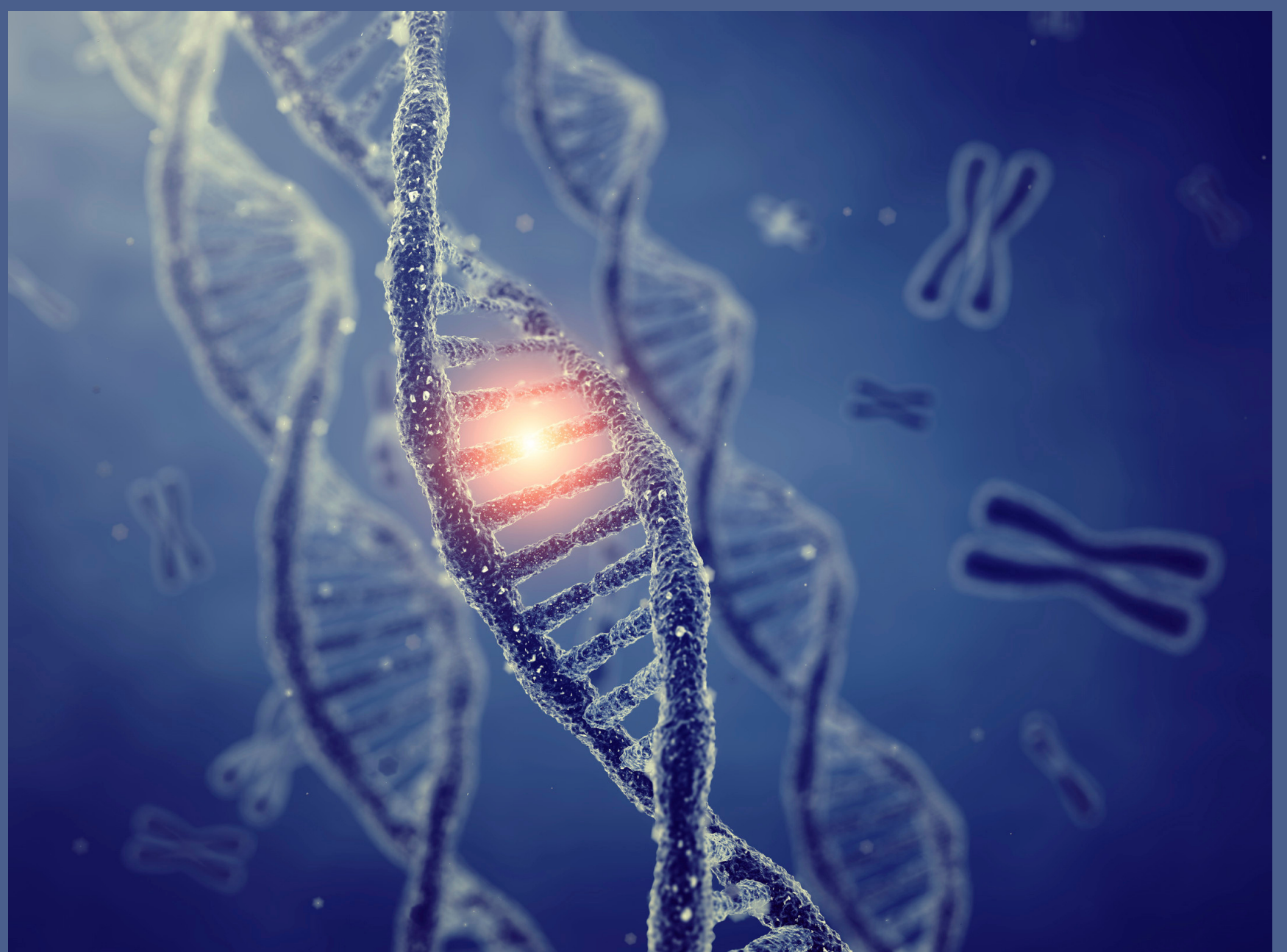
22 APRIL AT 2:30 P.M.  
ROOM A208 | POVO 1

CIBIO  
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seminar

# AHI1 MUTATIONS REVEAL A CONNECTION BETWEEN PRIMARY CILIUM AND CELL CYCLE PROGRESSION

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The use of cellular models obtained directly from cells of patients carrying genetic mutations that cause rare diseases makes it possible to study the molecular mechanisms underlying the disease in vitro. We obtained two fibroblast lines from two patients carrying homozygous mutations in the AHI1 gene, which causes Joubert's syndrome. We studied how AHI1 influences primary cilium formation and how elongation and resorption of the primary cilium influence cell cycle progression. We found pathological mechanisms affecting Joubert cells that could be used as targets to revert pathological phenotype.



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