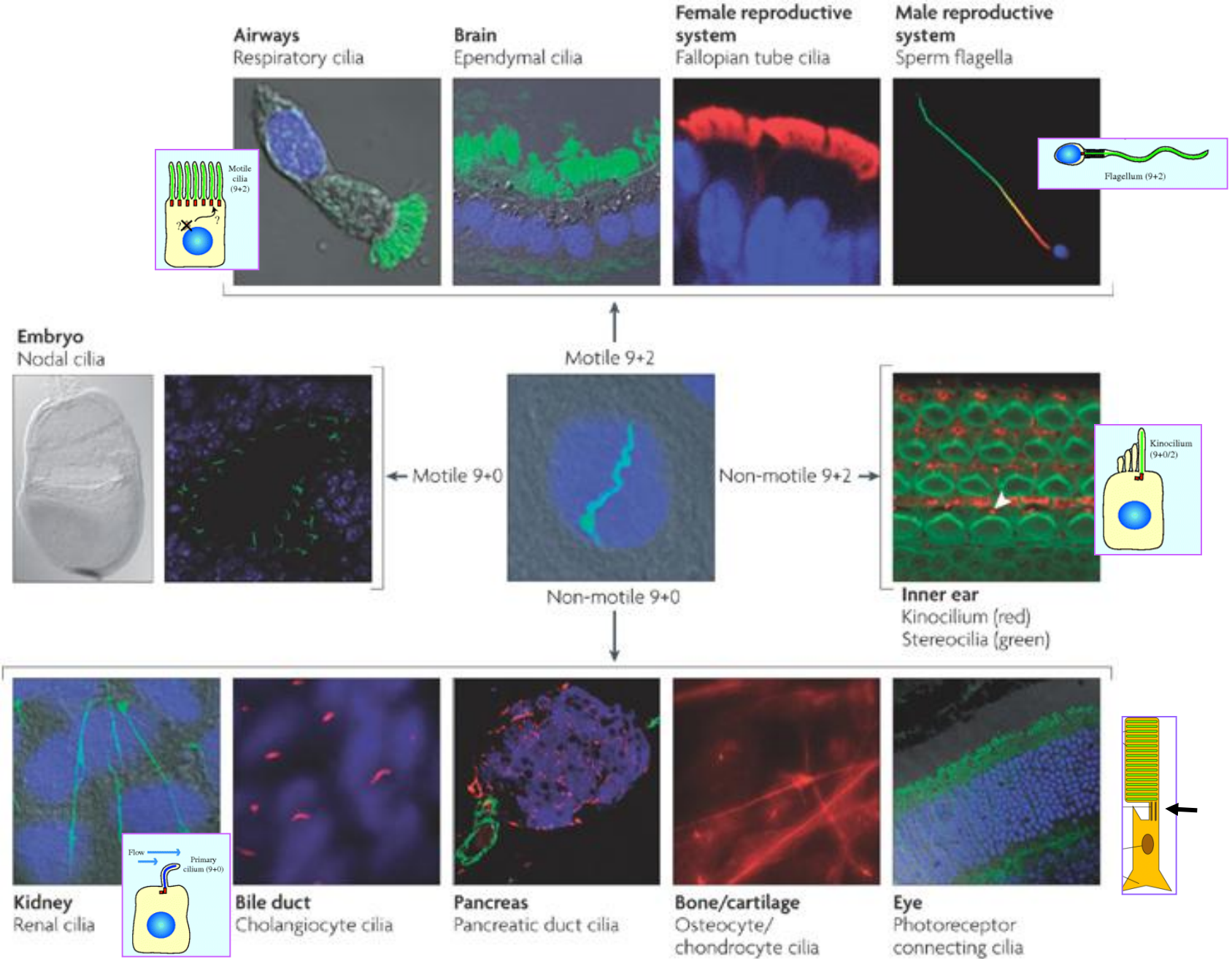


The Medical Impact of Cilia

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Major 'ciliopathy' phenotypes

	PKD	NPHP	MKKS	SLSN	EVC	JATD	OFD	ALMS	JS	BBS	MKS	PCD
CNS malformations					●	●	●	●	●	●	●	
Cystic kidney	●	●		●		●	●	●	●	●	●	●
Diabetes								●		●		
Gonadal malformations			●		●					●	●	
Heart disease			●		●			●		●		●
Hepatic dysfunction	●	●		●		●	●	●	●	●	●	
Mental retardation/Developmental delay					●	●	●		●	●	●	
Obesity								●		●		
Polydactyly			●		●	●	●		●	●	●	
Pulmonary dysfunction								●				●
Retinal degeneration				●		●		●	●	●		
Left-right asymmetry defects		●		●		●			●	●	●	●
Skeletal defects					●	●	●					

PKD, polycystic kidney disease; NPHP, nephronophthisis; MKKS, McKusick–Kaufman syndrome; SLSN, Senior–Løken syndrome; EVC, Ellis–van Creveld; JATD, Jeune asphyxiating thoracic dystrophy; OFD, orofaciodigital syndrome; ALMS, Alström syndrome; JS, Joubert syndrome/Cerebello–oculo–renal syndrome; BBS, Bardet–Biedl syndrome; MKS, Meckel–Gruber syndrome; CNS, Central nervous system.

Ciliopathy diseases

- >20 disorders. Defects in the function or structure of cilia
- Cilia:
 - Sensory antenna signalling responses to mechanical and chemical changes
 - Motile, transporting material across cells & moving cells through fluid
- Affect multiple systems, causing blindness, deafness, chronic respiratory infections, kidney disease, heart disease, infertility, obesity and diabetes
- Regarded as separate diseases, but share clinical and biological similarities
- Many of these conditions are rare (not PKD) and attract minimal funding for research, disease management and treatments
- As a group they have a significant health economic impact