

Table 1. Organ involvement in selected ciliopathies. Ciliary dyskinesia, indicates motile cilia structure/function defects. PCD, primary ciliary dyskinesia, RGMC, reduced generation of multiple motile cilia.

Ciliopathy	Primary affected organs	Symptoms associated with motile cilia and/or nodal cilia involvement (see text for gene details)
Primary ciliary dyskinesia	Respiratory, brain, heart, organ placement, sperm	PCD, RGMC
MORM syndrome	Brain, kidney, eyes	
Meckel-Gruber syndrome	Brain, kidney, skeleton	Situs inversus
Bardet-Biedl syndrome	Brain, kidney, skeleton, eyes	Situs inversus, ciliary dyskinesia reports
Oculocerebrorenal (Lowe) syndrome	Brain, kidney, skeleton, eyes	
Orofaciodigital syndrome	Brain, kidney, skeleton, liver	OFD1 – situs inversus, heart defects
Simpson-Golabi-Behmel syndrome, type 2	Various, respiratory	OFD1 – brain malformation, ciliary dyskinesia
COACH syndrome	Brain, renal	Situs inversus
Hydrolethalus syndrome	Brain, skeleton	Situs inversus
Pallister-Hall syndrome	Brain, skeleton	
Joubert syndrome	Brain, various	Situs inversus
Acrocallosal syndrome	Brain	
Holoprosencephaly	Brain	Situs inversus
Juvenile myoclonic epilepsy	Brain	
Medulloblastoma	Brain	
Stromme syndrome	Brain	
Cone-rod dystrophy	Eyes	Situs inversus, heart defects
Leber congenital amaurosis	Eyes	CEP290 – ciliary dyskinesia
Retinitis pigmentosa	Eyes	RPGR - respiratory defects, PCD
Senior-Løken syndrome	Kidney, eyes	Situs inversus
Nephronophthisis	Kidney	NPHP2 - situs inversus, ciliary dyskinesia
Polycystic kidney disease	Kidney	PKD2, IFT88 - situs inversus
Alström syndrome	Obesity, endocrine, eyes	Situs inversus
Cranioectodermal dysplasia (Sensenbrenner)	Skeleton, kidney, hepatic	Ciliary dyskinesia
Axial spondylometaphyseal dysplasia	Skeleton	
Carpenter syndrome	Skeleton	Situs inversus
Ellis-van Creveld syndrome	Skeleton	Situs inversus
Grieg cephalopolysyndactyly syndrome	Skeleton	
Jeune asphyxiating thoracic dystrophy	Skeleton	Situs inversus
McKusick-Kaufman syndrome	Skeleton	
Short-rib thoracic dystrophy	Skeleton	Situs inversus
Birt-Hogg-Dubé syndrome	Various	
Lethal congenital contracture syndrome	Various	
Spinocerebellar ataxia	Various	