

# DYNC2H1

## Short Rib Thoracic Dysplasia, with or without polydactyly

<b>Mode of Inheritance</b>	<ul style="list-style-type: none"><li>• Autosomal recessive</li><li>• Variable expressivity</li></ul>
<b>Renal Phenotype</b>	<ul style="list-style-type: none"><li>• Polycystic kidneys, renal dysplasia</li><li>• Age of onset: Early childhood</li></ul>
<b>Extra-renal Manifestations</b>	<ul style="list-style-type: none"><li>• Short stature</li><li>• Retinal dystrophy</li><li>• Cleft lip/palate</li><li>• Cardiac defects</li><li>• Restrictive lung disease</li><li>• Biliary dysplasia or cystic pancreas (rare)</li><li>• Intestinal malrotation, anal atresia</li><li>• Scoliosis and skeletal abnormalities (e.g. shortened long bones, brachydactyly)</li></ul>
<b>Pre-Transplant Management</b>	<ul style="list-style-type: none"><li>• Screening and management of extra-renal manifestations</li><li>• Optimization of respiratory status</li></ul>
<b>Transplant Considerations</b>	<ul style="list-style-type: none"><li>• Tailor immunosuppression given low risk of recurrence post-transplant</li></ul>
<b>Post-Transplant Management</b>	<ul style="list-style-type: none"><li>• Low risk of disease recurrence</li></ul>