

SALL1

Townes-Brocks Syndrome

Mode of Inheritance	<ul style="list-style-type: none">• Autosomal dominant• Variable expressivity
Renal Phenotype	<ul style="list-style-type: none">• Variable, including: hypodysplasia, multicystic dysplastic kidneys, and renal failure
Extra-renal Manifestations	<ul style="list-style-type: none">• Microcephaly• Sensorineural hearing loss• External ear malformations• Eye anomalies (coloboma, duane anomaly – rare)• Cardiac defects• Anorectal malformations (e.g. imperforate anus)• Hypospadias, cryptorchidism• Broad thumb, polydactyly, syndactyly
Pre-Transplant Management	<ul style="list-style-type: none">• Screening and management of extra-renal manifestations
Transplant Considerations	<ul style="list-style-type: none">• Careful screening of potential living related donors (inherited in an autosomal dominant manner)
Post-Transplant Management	<ul style="list-style-type: none">• Low risk of disease recurrence