4p13 Deletion
Wolf-Hirschhorn Syndrome

| Mode of Inheritance | • Primarily de novo deletions (87%)
• Variable expressivity |
<table>
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<td>Renal Phenotype</td>
<td>• Variable, including: vesicoureteral reflux, renal agenesis, cystic dysplasia, hypoplasia, horseshoe kidney, bladder extrophy</td>
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| Extra-renal Manifestations | • Failure to thrive
• Microcephaly
• Facial features: high forehead, hypertelorism, beaked nose, downturned mouth, micrognathia
• Cleft lip or palate
• Small bowel malrotation, reflux, absence of the gallbladder
• Genital anomalies (e.g. hypospadias, cryptorchidism, clitoral aplasia or hyperplasia, absent uterus/vagina)
• Scoliosis, polydactyly, fused vertebrae or ribs
• Developmental delay and hypotonia
• Hydrocephalus, corpus callosum abnormalities
• Seizures |
| Pre-Transplant Management | • Screening and management of extra-renal manifestations |
| Transplant Considerations | • Careful screening of potential living related donors (inherited in an autosomal dominant manner) |
| Post-Transplant Management | • Low risk of disease recurrence |

https://www.omim.org/entry/194190