

# The Genetics of Primary Vesico-Ureteric Reflux

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## Abstract

**Introduction:** Vesico-ureteric reflux (VUR), reflux of urine from the bladder into the ureter and towards the kidney, is an important cause of end-stage renal failure in both children and adults. Primary VUR is considered to be a result of a disruption of the normal anti-reflux mechanism of the ureterovesical junction (UVJ). VUR is common, occurring in approximately 1% to 2% of newborns in Caucasian populations. The aetiology of VUR is thought to involve a substantial genetic component, supported by the observation that VUR frequently occurs in multiple members of the same family. The purpose of this article is to review the literature supporting a genetic cause of VUR, and to draw together observations and make suggestions regarding differential diagnosis of VUR, which might help in future studies on the genetic aetiology of VUR. **Results:** A common theme arising was the notion that VUR may be caused by multiple genes in the population. However, any one individual with VUR may carry a single dominant mutant allele. Overall, progress has been made in mapping putative VUR loci in both humans and mice, although the mode(s) of inheritance and the exact nature of the underlying defect are still poorly understood. **Conclusions:** It is likely that over the next few years VUR genes will be mapped and, once identified, the challenge will be to understand how changes in the expression of these genes lead to the underlying defect in VUR.

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**Key words:** Kidney damage, Pyelonephritis, Reflux nephropathy, Ureter development, Urinary tract infection

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