Title: Noonan Syndrome GeneReview — Spatial distribution of the common PTPN11 variant c.922A>G in testes
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Note: The following information is provided by the authors listed above and has not been reviewed by GeneReviews staff.

Yoon et al [2013] examined the spatial distribution of the common PTPN11 variant c.922A>G in testes from 15 unaffected men and found that the pathogenic variants were not uniformly distributed across each testis as would be expected for a mutational hot spot but were highly clustered and showed an age-dependent germline mosaicism. It is suspected that mutated spermatogonial stem cells gain an advantage that allows them to increase in frequency. SHP-2 interacts with the transcriptional activator STAT3. Given STAT3's function in mouse spermatogonial stem cells, this interaction might explain the mutant's selective advantage by means of repression of stem cell differentiation signals.

References