Background: It is as a tenet in the field that most hypospadias cases are idiopathic. In spite of great research efforts, there is little information about the relationships between the genetic and environmental underpinnings of the condition to geographical space. Therefore, we aimed to determine the spatial distribution of cases within a well-defined geographic location.

Methods: The dataset for this study was produced by the Birth Defects Prevention and Surveillance System of the Department of Health of Puerto Rico (2007-10; n=279) and a clinical dataset from three participating institutions from 2007-13 (n=142). A population-based case-control study was conducted to estimate the potential effects of maternal, paternal, birth-related variables, and social risk factors for hypospadias. Two types of geographical information systems (GIS) methods (Anselin Local Moran’s I and Getis-Ord G) were used to determine the spatial distribution of hypospadias according to prevalence and severity.

Results: Gestational age (25 to 37 weeks), age of the mother (40 +), and being non-poor were found as risk factors for having a newborn with hypospadias as confirmed with univariate and multivariate analyses at 95% CI. A cluster of hypospadias cases was detected in the north-central region of Puerto Rico with both GIS methods (p < 0.05). Clusters of cases were also detected according to severity.

Conclusions: The clustering of hypospadias by prevalence and severity provides an opportunity to assess the underlying causes of the condition and their relationships with geographical space.