Elucidating the diagnostic odyssey of 22q11.2 deletion syndrome

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Individuals with 22q are often undiagnosed for years because this condition is not easily recognizable. This study analyzed the time it took for a diagnosis of 22q and reasons for delays across age and groups in Toronto and Philadelphia. Problems with the palate and heart were associated with shorter times to a 22q diagnosis. Non-European ancestry lead to longer times. There is a great need for education about 22q so that the condition is more widely known.