

Primary ciliary dyskinesia occurs in approximately 1 in 16,000 to 20,000 births. That translates to the incidence of Kartagener syndrome as 1 in 32,000 to 40,000 births. Primary ciliary dyskinesia is diagnosed definitively through examination of lung or sinus tissue obtained from a biopsy. Specific structural defects that are present in these tissues can be detected under an electron microscope. Early diagnosis is important in order to provide prophylactic treatment to prevent or decrease damage to the respiratory system from recurrent infections. Screening for levels of nasal nitric oxide (in patients over 5 years of age who can cooperate with palate closure maneuvers) is helpful to identify individuals who may have PCD and should proceed with a biopsy. Currently, mutations in 32 genes are known to be associated with PCD. These do not account for all cases of PCD and hence more PCD genes are yet to be identified. PCD clinical genetic testing is available for some of the 32 genes associated with PCD by the commercial laboratories and new genes are being added to their panels periodically.