Title: *LRRK2* Parkinson Disease *GeneReview* – Prevalence of Variants in Different Populations
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Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

p.Gly2019Ser

- Approximately 30%-34% of simplex PD and up to 41% of familial PD in the North African Berber population (Morocco, Algeria, and Tunisia) [Hulihan et al 2008, Lesage et al 2008]
- Approximately 18% of PD in Ashkenazi Jews in New York, US (13% of simplex cases and 30% of familial cases) [Ozelius et al 2006]
- Approximately 15% of PD in Tel Aviv, Israel (10.6% of simplex Ashkenazi Jewish cases and 26% of familial cases [Orr-Utreger et al 2007]
- 3.7%-4.9% simplex and 9.1%-16.1% familial PD in Portugal [Bras et al 2005, Ferreira et al 2007]
- 0.9%-2% simplex PD and 4%-5.1% familial PD in Italy [Goldwurm et al 2005, Healy et al 2008] (See also p.Arg1441Cys below.)
- 2.7%-8.7% in northern Spain [Mata et al 2006, Sierra et al 2011];1.7% of simplex PD in southern Spain [Gao et al 2009] (See also p.Arg1441Gly below.)
- Approximately 0.5% of simplex PD and 2%-6% of familial PD in the US [Deng et al 2005, Farrer et al 2005, Nichols et al 2005, Kay et al 2006]
- Approximately 0.5% of simplex PD in the UK [Williams-Gray et al 2006]
- The p.Gly2019Ser variant was reported in two out of eight individuals with PD from Puerto Rico who were screened for this specific variant [Saunders-Pullman 2011]
- Approximately 2% in families from central Norway [Aasly et al 2005]
- Approximately 0.76% simplex PD and 8.7% familial PD in Brazil [Pimentel et al 2008]
- 0.4% of simplex PD in Japan [Zabetian et al 2006]

Note: p.Gly2019Ser was reported in a Japanese individual with PD in another study [Tomiyama et al 2006]. In that study, however, the frequency of p.Gly2019Ser was reported in a pooled Asian sample and the frequency among Japanese individuals with PD was not specified.

The **p.Gly2019Ser** variant is not common in the following populations:

- A single p.Gly2019Ser variant was detected in a screen of 1,012 individuals with PD in India [Punia et al 2006]. This pathogenic variant has not been

observed in 326 individuals with PD of South Indian ancestry [Vijayan et al 2011, Vishwanathan et al 2012].

- p.Gly2019Ser was observed in a single individual of Han Chinese ancestry, but was absent in screens of more than 2,700 individuals with PD of Chinese ancestry [Lu et al 2005, Tan et al 2006, Tomiyama et al 2006, Tan et al 2010, Zhang et al 2018].
- The p.Gly2019Ser pathogenic variant has not been observed in screens of 183 Nigerian Africans with PD nor in healthy controls [Okubadejo et al 2008, Okubadejo et al 2018].

p.Arg1441Gly

- Is uncommon outside Spain, as only a single affected individual with this variant has been reported outside Spain, in a Japanese family [Hatano et al 2014]
- Is present in approximately 8% of individuals with PD from the Basque community in northern Spain due to a founder effect [Paisan-Ruiz et al 2004, Mata et al 2005, Deng et al 2006, Simón-Sánchez et al 2006, González-Fernández et al 2007, Mata et al 2009]

p.Arg1441Cys

- Has been reported in individuals of Belgian, Italian, German, Irish, Chinese, and Singaporean ancestry [Zimprich et al 2004, Haugarvoll et al 2008, Peng et al 2017], but is likely uncommon outside southern Italy and Belgium [Pankratz et al 2006, Peng et al 2017]
- Is more common than p.Gly2019S in individuals of Southern Italian ancestry (particularly from Campania where Naples is located). The prevalence of this variant has been found to be 2.5%-4.7% in individuals with PD from Campania [Criscuolo et al 2011, De Rosa et al 2014].
- May be a founder variant in Belgium, where 2% of individuals with PD have this variant [Nuytemans et al 2006]

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