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LETTER TO THE EDITOR

Application of long-range polymerase chain reaction in the diagnosis of X-linked dystonia–parkinsonism

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Sir,

X-linked dystonia–parkinsonism (XDP, DYT3, also referred to as "Lubag") is a neurodegenerative disorder characterized by a unique combination of parkinsonism and dystonia [1]. The insertion of short interspersed nuclear element, variable number of tandem repeats, and Alu composite (SVA) retrotransposon has been identified in intron 32 of the TATA-binding protein-associated factor 1 gene (TAF1), which is mapped within the haploblock associated with XDP [2]. Several disease-specific single-nucleotide changes (DSCs) and 48-bp deletion polymorphism have also been mapped within the haploblock [3]. The DSC3, located at the TAF1/DYT3 multiple transcript system, has been investigated and shown to be associated with XDP [4]. TAF1 is a component of the transcription initiation factor TFIID which plays a central role in mediating promoter responses to various activators and repressors [5]. DSC3 containing transcripts as well as alterations of TAF1 splice variants would affect the transcription of several genes, eventually leading to neurodegeneration [2, 4]. Genetic testing for XDP has been performed using Southern analysis for SVA retrotransposon or direct polymerase chain reaction

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