Rapid-onset dystonia parkinsonism (RDP) is a rare disorder characterized by abrupt onset of dystonia and parkinsonism. The condition is the result of mutations in \textit{ATP1A3}. Careful characterization of neurological manifestations associated with \textit{ATP1A3} mutations, which can cause RDP and alternating hemiplegia of childhood (AHC), is important to understand how specific mutations can lead to different presentations. This case of RDP is notable because (1) the patient harbored an \textit{ATP1A3} variant at the c.2267G>A site, resulting in a non-synonymous p.R756H mutation, which has not been reported in typical adolescent-onset RDP, and (2) reports of Asian cases are very rare and this is the first patient of pure Chinese descent.

Our patient, of pure Southern Chinese ancestry, was previously healthy until 9 May 2002 (at the age of 10 years), when she became clumsy. This was preceded by a self-limited febrile episode a week earlier. The next day, she had dysarthria, drooling, unsteady gait, and poor handwriting. This progressed over 10 days to a state of being unable to speak, swallow, or walk. She was recognized to have a severe dystonic syndrome when assessed by a pediatric neurologist 3 weeks later. By this time, she had made some recovery and was able to walk with assistance and swallow. Investigations were unremarkable, including routine blood tests, cerebrospinal fluid analysis, brain MRI, and EEG. Slit-lamp examination was negative for Kayser-Fleischer rings. She was diagnosed with postviral encephalitis and given levodopa/carbidopa (100/25 mg, 2 tablets three times daily), but this was ineffective.

The patient’s DNA was analyzed for \textit{ATP1A3} mutations, and she was found to have a mutation in exon 17 at cDNA position c.2267G>A (NM_152296.4), resulting in a p.R756H amino acid substitution (NP_689509; laboratory of L.J.O.). Neither of the patient’s parents had this mutation. DNA profiling was performed using short tandem repeat (STR) analysis at 15 STR loci (D8S1179, D21S11, D7S820, CSF1PO, D3S1358, TH01, D13S317, D16S539, D2S1338, D19S433, vWA, TPOX, D18S51, D5S818, and FGA) by an accredited national forensics laboratory; this confirmed that the patient was the biological child of the parents in question.

Benzhexol 2 mg TID subjectively reduced limb stiffness and improved speech, but caused sleepiness and dizziness and was stopped. Her condition has remained stable (see accompanying Video 1 from November 2013), with a Burke-Fahn-Marsden Dystonia Rating Scale Movement Scale score of 67, a Disability Scale score of 13; an RDP Severity Scale score of 4; and the following UPDRS scores (largely felt to be a result of the severe dystonia; see Video 1): Total of 55.5, Parts I of 0, II of 42.5, and III of 35.5. She had a Motor Score of 29/30.

To our knowledge, the \textit{ATP1A3} p.R756H mutation has not been reported in typical adolescent-onset RDP. This mutation...
has been reported only once previously, in a novel infant-variant (i.e., very young onset) case of RDP with atypical clinical features, who inherited the mutation from her father who had only minor neurological symptoms.\(^4\) These cases thus illustrate that the same \(ATP1A3\) mutation can result in vastly different clinical outcomes. We believe that this is also the first case of RDP to be reported in a patient of pure Chinese ethnicity; the only other Asian cases in the literature were a Korean man and a child of mixed Caucasian-Chinese parentage\(^5,6\) (a cohort of Chinese patients with AHC and \(ATP1A3\) mutations was reported on recently, but none exhibited an RDP phenotype).\(^7\) Our case resulted from a de novo mutation, highlighting the need to consider a diagnosis of RDP even without a family history of similar illness.\(^1\)

**Author Roles**


A.H.T.: 1B, 1C, 3A, 3B
L.J.O.: 1C, 3B
A.B.: 1A, 1C, 3B
A.E.L.: 1C, 3B
A.A.-A.: 1C, 3B
C.T.T.: 1C, 3B
S.-Y.L.: 1A, 1B, 1C, 3A, 3B

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**Disclosures**

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**References**


**Supporting Information**

A video accompanying this article is available in the supporting information here.

**Video 1.** Severe dystonic posturing of the limbs, neck (ante-collis), and trunk (especially upon walking). Speech is severely slurred with jaw-opening dystonia. The patient could stand up independently from a sitting position, but was very slow to do so. Her slowness of movements was considered to be a result of the severe dystonia.

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**CASE REPORT**

**RDP in a Chinese Girl**


