A mother and son with Noonan syndrome resulting from a *PTPN11* mutation

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Letter to the Editor

Recently, we read a report about Noonan syndrome entitled "A mother and son with Noonan syndrome resulting from a *PTPN11* mutation: first report of molecularly proven cases from Turkey" published in the Turkish Journal of Pediatrics (2010; 52: 321-324). The authors reported a mother and son with Noonan syndrome (NS) whose molecular analysis showed an A923G mutation in exon 8 of the *PTPN11* gene. It was noted that this is the first report of molecularly proven cases from Turkey. However, before that, there were two studies about NS patients and their *PTPN11* gene analysis results in both a national journal and two Congresses in Turkey[1-3]. Furthermore, the same A923G mutation and its clinical picture was previously reported in a national journal[3].

In 2006, we studied 12 clinically diagnosed patients with NS whose molecular analysis was performed in the Center for Human Genetics, University of Leuven, Belgium. Blood samples were analyzed for mutations of the *PTPN11* gene. The results of our study were presented as an oral presentation in a national Turkish Congress[1]. Later, we studied the clinical and hematologic features of NS patients with *PTPN11* mutation in a larger series. We published our results in another national Turkish Pediatrics Congress Book and an international journal[3,4].

In 2009, Altunoğlu et al.[3] reported the clinical data of 35 patients and mutation analysis results of the *PTPN11* gene, and also other responsible genes of NS, including *SOS1, KRAS* and *RAF1*. The genotype-phenotype correlation was investigated in that study. Their one NS patient had died after the operation for grade II astrocytoma. This patient had the same A923G mutation in the *PTPN11* gene.

The associations of myeloproliferative disorders, bleeding diathesis and tumor development are well-known features of NS[4-6]. Thus, the presented NS patient with A923G mutation should be followed-up for malignancy and hematologic findings.

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