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EXCLUSIVE

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conclusion: there was evidence that pth was positively correlated with cardiac iron in this study, confirming previous reports. these patients had a high prevalence of vitamin d deficiency. these patients should receive evaluation for cardiac iron to prevent sequelae. this study also provided evidence for pth as a simple and quick laboratory parameter for evaluation of patients with thalassemia. aim and method: patients with noonan syndrome (ns) have a distinctive facial appearance, showing short stature, round face, large forehead, hypertelorism, and short, webbed neck. they have a typical congenital heart disease, including hypertrophic cardiomyopathy, atrial septal defects and pulmonary arteriovenous malformations. moreover, platelet-derived growth factor receptor α (pdgfr α) and fmsH2 mutations were recently identified as the cause of ns. pdgfr α -deficient cells showed abnormal cell cycle progression and development of cells with enlarged and/or ruffled cell membrane. both non-transformed and transformed cells have a decreased growth rate when they have reduced levels of fmsH2. because these mutations are inherited as autosomal dominant disorders, it is possible to make future predictions about the presence of phenotype. however, a few cases of mutation-negative ns have been reported. case: we present the case of a 14-year-old female patient with a 4-year history of mild to moderate exercise-associated fatigue, abnormal palpitations, and subclinical hypothyroidism. she had developmental delay from early infancy. after puberty, she had easy fatigability without any other remarkable disease. she had normal development at elementary school but had mild developmental delay. she had no history of

medication and eating disorder, except for excessive exercise. her height was 155 cm and body weight was 60 kg. she was pale but showed no dysmorphism except for a round face, hypertelorism, short neck, and low-set ears.

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