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Editorial

Comprehensive care for thalassemia: not just the treatment of anemia

Thalassemia syndromes are the commonest monogenic disease worldwide, with at least 60,000 severely affected newborns every year [1]. In Thailand where thalassemia syndromes are highly prevalent, approximately 600,000 Thais suffer from thalassemia diseases [2]. The life expectancy of patients with thalassemia in the recent decade has remarkably improved because of better treatment strategies such as transfusion policy, adequate iron chelation therapy, and substantial improvement in hematopoietic stem cell transplantation [3-6]. Although cardiac failure remains the major cause of death, several complications are increasingly recognized in long-term survivors. However, the data of these long-term complications in Thai thalassemia patients are lacking.

In this issue of *Asian Biomedicine*, two studies address common, but yet usually neglected complications in the general care for thalassemia patients. Nakhakes and colleagues report the prevalence and risk factors for osteoporosis in 190 adults with thalassemia intermedia and thalassemia minor [7], while Siripunthana and colleagues investigated testicular function in 28 pediatric patients with regular blood transfusions to treat their thalassemia [8].

Of 190 adults including those with 138 thalassemia intermedia and 52 thalassemia minor, the prevalence of osteoporosis is 11.6%. The prevalence and the severity of osteoporosis are more pronounced in patients from the subgroup with thalassemia intermedia. Although the authors fail to identify significant risk factors associated with osteoporosis in this cohort, they showed that thalassemia patients have a substantial high prevalence of vitamin D deficiency comparing with the general Thai population. In addition, the vast majority of patients have inadequate intakes of calcium [7]. Whether these findings are associated with an increased risk of osteoporosis in thalassemia patients remains to be determined. The authors recommend that routine nutrition assessment and nutrition support should be integrated in comprehensive care of patients with thalassemia. The prevalence of osteoporosis among thalassemia intermedia in this study is much lower than that found in the previous study by Taher and colleagues (13.78% vs. 22.9%) [7, 9]. The great variety of genotypic determinants, patient selection, and treatment strategies might explain this significant difference. In the large cohort from Mediterranean Europe and the Middle East, age >35 years, female sex, splenectomy, and regular blood transfusion were associated with an increased risk for osteoporosis, while iron chelation and hydroxyurea treatment are protective factors [9]. Nevertheless, the pathogenesis of osteoporosis in thalassemia disease remains poorly understood.

Of 28 pediatric patients with β-thalassemia disease receiving regular transfusion, the testosterone levels were lower than those of normal controls, especially in patients at Tanner stage IV–V, although the puberty of patients was not delayed. The values of serum follicle-stimulating hormone and luteinizing hormone were not significantly different from those of the control group [8]. These findings suggest preserved Sertoli cell function, but subclinical Leydig cell dysfunction at the late pubertal period in patients with β-thalassemia receiving regular transfusion and adequate iron chelation. In addition, this study encourages the regular assessment of growth, development, and testicular function in pediatric patients with thalassemia to detect abnormalities early and initiate early treatment. The prevalence of hypogonadism and delayed puberty in Thai pediatric patients is substantially lower than that in previous reports [10-12]. This is likely to be because of the different population studied. Indeed, the majority of patients in the current study were β-thalassemia/HbE, a genotype that usually leads to the thalassemia intermedia phenotype [13-14]. The long-term complications observed between patients with thalassemia intermedia and thalassemia major are remarkably different [15]. Therefore, the authors should conduct further studies to evaluate testicular function in the thalassemia major group.

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In general practice, physicians usually evaluate treatment using hemoglobin and serum ferritin levels, while other complications are rarely screened for. The two current studies in this journal should increase physicians' awareness about other common long-term complications in thalassemia diseases. However, the mechanisms underlying these abnormalities remain to be elucidated. Moreover, when these complications have been detected, the interventions attempting to either reverse or improve outcomes are usually unsuccessful. Therefore, the prevention of new cases is much more cost-effective. The effective prevention and control program for thalassemia has been successful in many countries including Cyprus, Italy, and Greece [16]. The keys of success are public awareness, education and nationwide accessibility to medical service. Although a national comprehensive prevention program for thalassemia has been established for more than a decade in Thailand [17], the outcomes are apparently unsatisfactory. The national health policy and the expenditure on public health should primarily focus on establishing an effective national program to prevent new affected cases.

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