CASE REPORT

Orthodontic Treatment of a Child with Hereditary Spherocytosis

JURANDIR ANTONIO BARBOSA, DDS, MS
PAULO ROBERTO FERREIRA LOPES, DDS
ELIANA MOREIRA BARBOSA LOPES, DDS

Hereditary spherocytosis (HS) is a common anemia with a heterogeneous mode of inheritance.[1,2] In northern Europeans, the prevalence of HS has been estimated at 1 in 5,000 births,[3] but this figure may be low considering recent data on the asymptomatic carrier state of the HS gene.[4] The disease has been identified in South African black populations, although its prevalence in these ethnic groups is unknown.[5-8] HS is the most common hemolytic anemia of congenital origin among Japanese.[9]

Two factors are involved in HS pathophysiology: a structural defect in the membranes of red blood cells and an intact spleen that selectively retains the abnormal cells, leading eventually to red cell destruction.[10,11] The clinical expression is highly variable, ranging from asymptomatic to severe, life-threatening hemolytic anemia. In most cases, the primary indicators include mild anemia, acholuric jaundice, splenomegaly, and a family history of the disease.[12]

Severe hemolytic anemia can lead to increased erythroblastic activity, which tends to expand the erythroid compartment in the bone marrow.[12,13] This marrow hyperplasia produces an enlargement of the diploe space in the frontal, parietal, and occipital regions of the skull, manifested as a prominent malar bone, an oversize nasal base, and a protrusive maxillary anterior region. Abnormalities in the mandible are usually more minor, including changes in the trabecular pattern, enlargement of the mandibular marrow space, and thickening of the lamina dura.[13-18]

HS cases with severe hemolytic anemia require a transfusion program with folic acid supplementation. If a splenectomy is performed, the erythrocytes will not be affected, but the destruction of red cells in the spleen may be slowed.[19] Most patients show a significant clinical improvement. Although there is always a risk of infection with surgery, the rate of morbidity is low if it is performed by an experienced surgeon.[20-23] Because of the increased susceptibility of young children to infections, a splenectomy is usually delayed until at least 6 years of age.[22-27]

This article describes a rare case in which an HS patient presented with typical anatomical signs of hemolytic anemia.

Dr. Barbosa is Graduate Program Director and Drs. Paulo Lopes and Eliana Lopes are Professors, Department of Orthodontics, Associação dos Cirurgiões Dentistas de Campinas, Campinas, São Paulo, Brazil. All of the authors are Professors, Department of Orthodontics, Centro de Pesquisas Odontologicas São Leopoldo Mandic, Campinas, São Paulo, Brasil, and in the private practice of orthodontics in São Paulo. Contact Dr. Paulo Lopes at Rua Duque de Caxias, 780/conj. 83, Centro, Campinas, SP 13015-311, Brazil; e-mail: elianapaulo @uol.com.br.


Dr. Barbosa


Dr. Paulo Lopes


Dr. Eliana Lopes

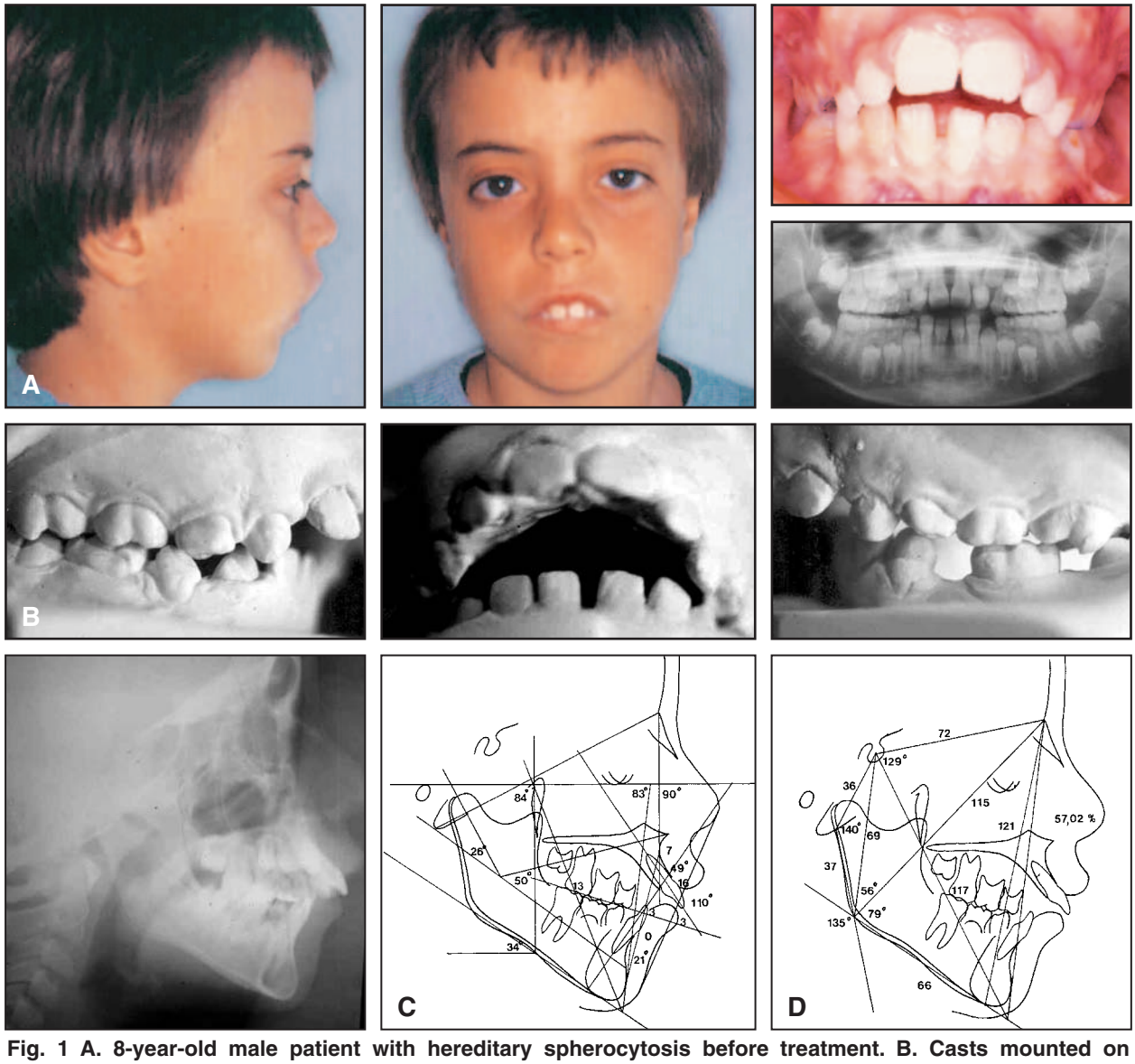

Fig. 1 A. 8-year-old male patient with hereditary spherocytosis before treatment. B. Casts mounted on Panadent* articulator. C. Ricketts cephalometric analysis. D. Björk-Jarabak cephalometric analysis.

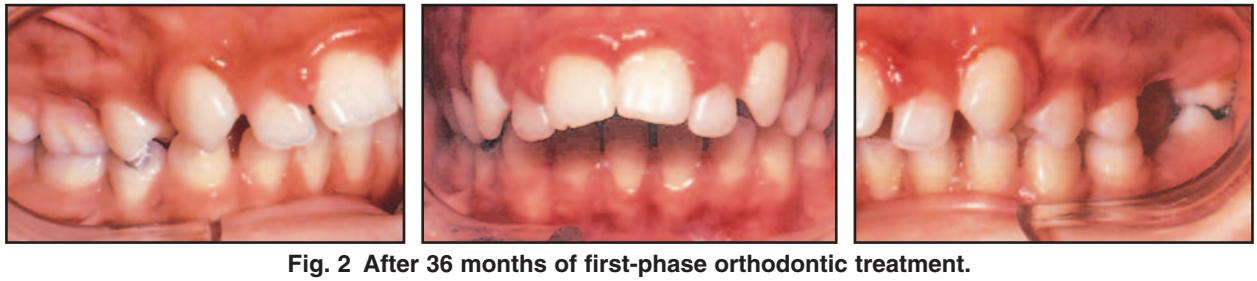

Fig. 2 After 36 months of first-phase orthodontic treatment.

Diagnosis

An 8-year-old boy sought orthodontic treatment because of excessive exposure of the maxillary incisors. His mother informed us that he was a carrier of HS, which was diagnosed when he was 2 months old. Since that time, he had been receiving periodic blood transfusions and taking folic acid daily.

Clinical examination revealed a convex profile, a closed nasolabial angle, and excessive protrusion and exposure of the maxillary anterior teeth (Fig. 1). The patient was in the mixed dentition, with a Class II, divi-

*Panadent Corp., 22573 Barton Road, Grand Terrace, CA 92313.

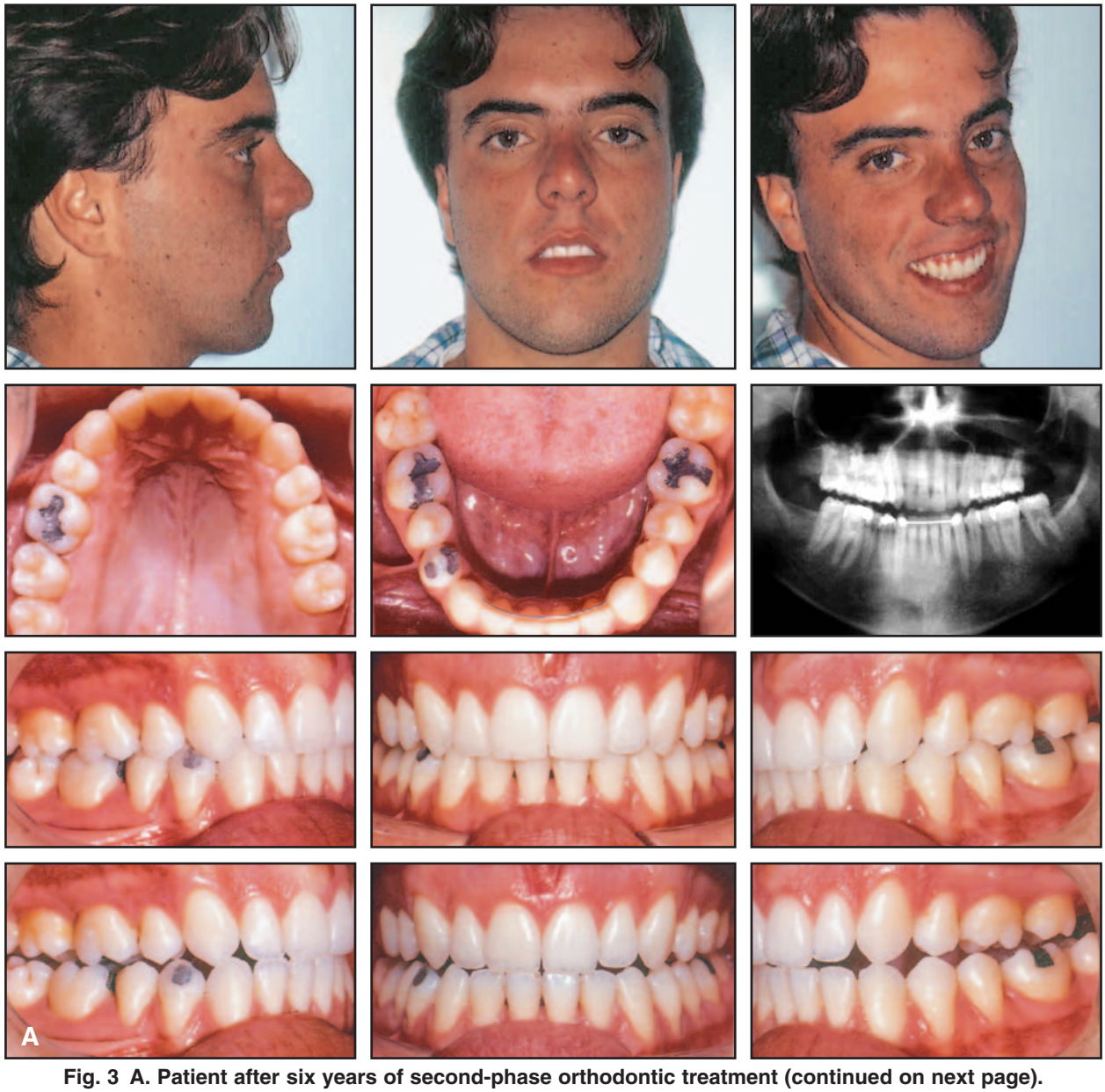

Fig. 3 A. Patient after six years of second-phase orthodontic treatment (continued on next page).

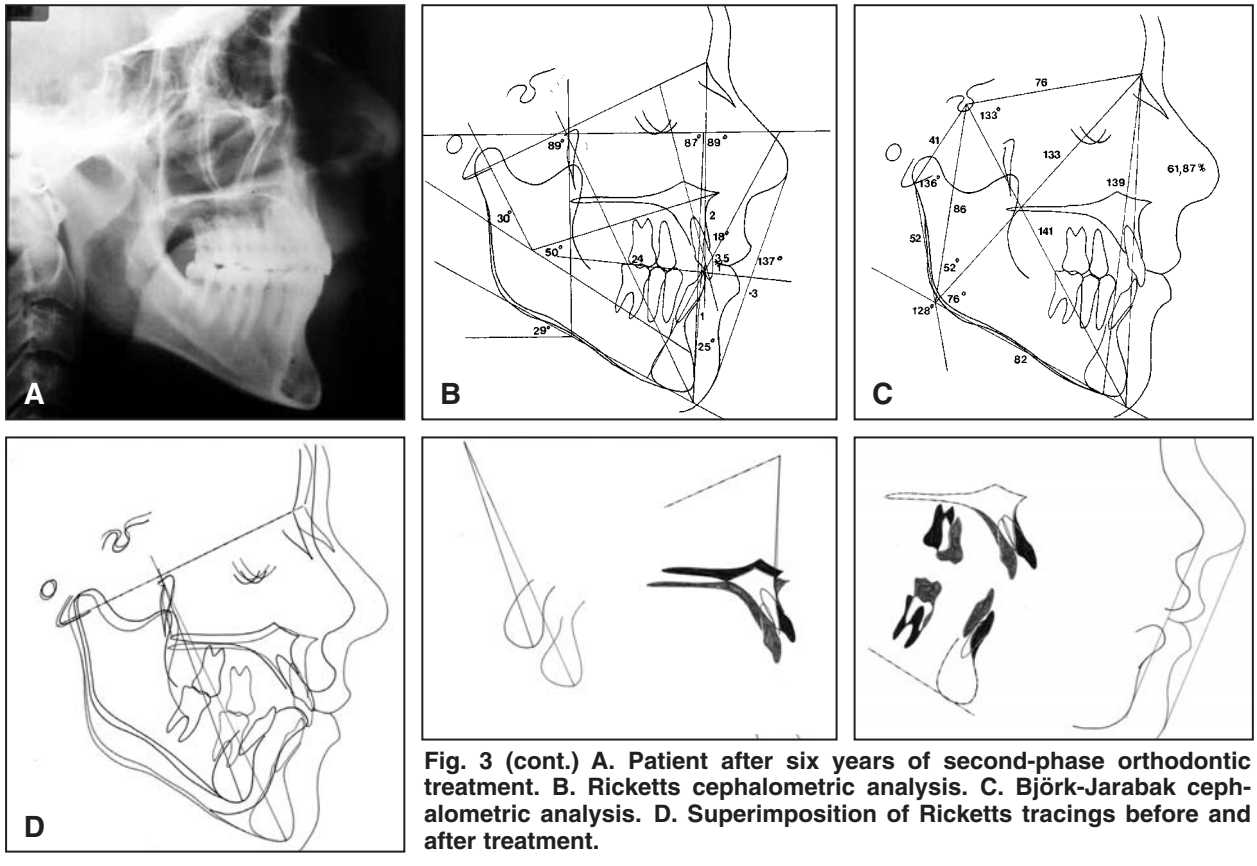

Fig. 3 (cont.) A. Patient after six years of second-phase orthodontic treatment. B. Ricketts cephalometric analysis. C. Björk-Jarabak cephalometric analysis. D. Superimposition of Ricketts tracings before and after treatment.

sion 1 malocclusion and a 3mm anterior open bite. He demonstrated generalized gingival inflammation and an absence of lip seal at rest.

The radiographs showed a migration of the mandibular left second bicuspid, trabecular bone alterations, enlargement of the mandibular marrow space, and thickening of the lamina dura and cortical frontal bone—all typical of HS. Cephalometrically, the patient had a dolichofacial pattern, a convex profile, a closed nasolabial angle, maxillary protrusion, and severe retrognathism due to a short ramus and mandible.

Treatment

Orthodontic treatment was begun with a high-pull headgear to correct the Class II malocclusion and the maxillary incisor protrusion. This stage was completed in 36 months (Fig. 2).

At age 12, when the permanent dentition had completely erupted, the hematologist ruled out extraction of the maxillary first premolars. A second phase of orthodontic treatment was initiated to correct the remaining tooth malalignments.

At age 13, the patient developed significant splenomegaly, and a splenectomy was performed. His blood transfusions and daily folic acid administration were stopped. The patient showed enough clinical improvement in six months that the hematologist authorized the premolar extractions.

The maxillary anterior segment was retracted to correct the overjet. After six years of second-phase treatment, final records demonstrated an improvement in the patient's occlusion, facial esthetics, and maxillomandibular function (Fig. 3).

The results have remained stable during a seven-year follow-up period.

Discussion

This HS patient's severe hemolytic anemia had produced facial and skeletal alterations that led to an extreme malocclusion. His therapy required constant coordination between the orthodontist and hematologist. Although the early orthodontic treatment produced little improvement, the patient showed a favorable response after a splenectomy was performed.

Orthodontists should be alert to the signs of HS and hemolytic anemia. If a splenectomy is considered as soon as possible, adverse facial and skeletal development may be prevented.

REFERENCES

1. Vanlair, C.F. and Masius, J.B.: De la microcythemie, Bull. R. Acad. Med. Belg. 5:515, 1871.
2. Bassères, D.S.; Duarte, A.S.; Hassoun, H.; Costa, F.F.; and Saad, S.T.: Beta-Spectrin S(ta) Barbara: A novel frameshift mutation in hereditary spherocytosis associated with detectable levels of mRNA and a germ cell line mosaicism, Br. J. Haematol. 115:347-253, 2001.
3. Morton, N.E.; MacKinney, A.A.; Kosower, N.; Schilling, R.F.; and Gray, M.P.: Genetics of spherocytosis, Am. J. Hum. Genet. 14:170-184, 1962.
4. Agre, P.; Asimos, A.; Casella, J.F.; and McMillan, C.: Inheritance pattern and clinical response to splenectomy as a reflection of erythrocyte spectrin deficiency in hereditary spherocytosis, N. Engl. J. Med. 315:1579-1583, 1986.
5. Nozawa, Y.; Hoguchi, T.; and Fukushima, H.: Erythrocyte membrane hereditary spherocytosis: Alterations in surface ultrastructure and membrane proteins as inferred by scanning electron microscopy and SDS on gel electrophoresis, Clin. Chim. Acta 55:81, 1974.
6. Hayashi, S.; Koomoto, R.; Yano, A.; Ishigami, S.; and Tsujino, G.: Abnormality in a specific protein of the erythrocyte membrane in hereditary spherocytosis, Biochem. Biophys. Res. Commun. 57:1038-1044, 1974.
7. Kline, A.H. and Holman, G.H.: Hereditary spherocytosis in the Negro, AMA J. Dis. Child. 94:609-615, 1957.
8. Metz, J.: Hereditary spherocytosis in the Bantu, S. Afr. Med. J. 33:1034-1036, 1959.
9. Yawata, Y.; Kanzaki, A.; Yawata, A.; Doerfler, W.; Ozcan, R.; Eber, S.W.: Characteristic features of the genotype and phenotype of hereditary spherocytosis in the Japanese population, Int. J. Hematol. 71:118-135, 2000.
10. Dacie, J.V. and Mollison, P.L.: Survival of normal erythrocytes after transfusion to patients with familial haemolytic anaemia (acholuric jaundice), Lancet 1:550, 1943.
11. Young, L.E.; Platzer, R.F.; Ervin, D.M.; and Izzo, M.J.: Hereditary spherocytosis, II. Observations on the role of the spleen, Blood 6:1099-1113, 1951.
12. Palek, J.: Hereditary spherocytosis, in *Hematology*, ed. W.J. Williams, E. Beutler, A.J. Erslev, and M.A. Lichtman, McGraw-Hill, New York, 1990, pp. 558-569.
13. Moseley, J.E.: Skeletal changes in the anemias, Semin. Roentgenol. 9:169-184, 1974.
14. Bellini, F.: Radiological pictures of skeletal changes in infantile hematologic diseases, Minerva Med. 64:2875-2883, 1973.
15. Marcinski, A.: Radiological changes in the skeletal system in cases of congenital hemolytic anemia, Pol. Przegl. Radiol. Med. Nukl. 30:461-470, 1966.
16. Okuda, K. and Yoshimura, K.: Case of hereditary spherocytosis with marked x-ray changes in the skull, Naika 16:960-963, 1965.
17. Sandoval, J.C.G.; Macías, F.C.; Macías, R.C.; Levy, M.A.M.; and Cedillo, J.A.S.: Hereditary spherocytosis in Baja California Sur, Mexico: Familial information and clinical aspects, Rev. Fac. Med. UNAM 32:19-24, 1990.
18. Tunaci, M.; Tunaci, A.; Engin, G.; Ozkorkmaz, B.; Dincol, G.; Acunas, G.; and Acunas, B.: Imaging features of thalassemia, Eur. Radiol. 9:1804-1809, 1999.
19. Chapman, R.G. and McDonald, L.L.: Red cell life span after splenectomy in hereditary spherocytosis, J. Clin. Invest. 47:2263-2267, 1968.
20. Van Wyck, D.B.: Overwhelming postsplenectomy infection: The clinical syndrome, Lymphol. 16:107-114, 1983.
21. Dawson, A.A.; Jones, P.F.; and King, D.J.: Splenectomy in the management of haematological disease, Br. J. Surg. 74:353-357, 1987.
22. Bridgen, M.L.: Postsplenectomy sepsis syndrome: How to identify and manage patients at risk, Postgrad. Med. 77:215-218, 1985.
23. Lux, S.E.: Disorder of the red cell membrane, in *Hematology of Infancy and Childhood*, vol. 1, ed. D.G. Nathan and F.A. Oski, Saunders, Philadelphia, 1987, pp. 489-490.
24. Lawrie, G.M. and Ham, J.M.: The surgical treatment of hereditary spherocytosis, Surg. Gynecol. Obstet. 139:208-210, 1974.
25. Rutkow, I.M.: Twenty years of splenectomy for hereditary spherocytosis, Arch. Surg. 116:306-308, 1981.
26. Baccarani, U.; Terrosu, G.; Donini, A.; Zaja, F.; Bresadola, F.; and Baccarani, M.: Splenectomy in hematology: Current practice and new perspectives, Haematol. 84:431-436, 1999.
27. Bolton-Maggs, P.H.: The diagnosis and management of hereditary spherocytosis, Baillieres Best Pract. Res. Clin. Haematol. 13:327-342, 2000.