Letters to the Editor

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Chediak-Higashi Syndrome (CHS): Report on Two Female Saudi Siblings

To the Editor: We read with interest the article of Al-Nasser et al.¹ on Chediak-Higashi syndrome (CHS) and share our experience of two Saudi female siblings.

Case 1. This patient was three years old in August 1991 when she was diagnosed as having CHS. Six months earlier, her one-year-old sister had been diagnosed as having CHS. Their parents are first cousins because their maternal and paternal grandmothers are full sisters.

This patient presented with a history of recurrent febrile illness, having been hospitalized several times in the past for bronchopneumonia. In addition, she had generalized hypopigmented and hyperpigmented hair and skin with freckles on her face, depigmented irides, enlarged liver and spleen that were 6 cm and 9 cm below the respective costal margins, moderate anemia and large intracytoplasmic granules in her leukocytes. Her CBC showed a Hb of 7.4 g/dL, WBC $4.8 \times 10^9/L$ and platelets $53 \times 10^9/L$. Her hemoglobin electrophoresis was 100% for HbA while her G6PD activity was normal. The chest radiograph revealed a right upper lobe consolidation and was treated with a combination of parenteral ampicillin and gentamicin as well as oral ascorbic acid for 10 days. Her clinical and hematological profiles improved before she was discharged home.

Nine months later she was admitted in the accelerated phase of CHS with cough, fever, generalized lymphadenopathy, hepatosplenomegaly and pancytopenia that was accompanied by severe anemia (Hb 4.3 g/dL) and bleeding (platelets $20 \times 10^9/L$). She was treated with acyclovir 15 mg/kg intravenously (IV) q. 8 h. and oral prednisolone 2 mg/kg/day in three divided doses for two weeks. She was also transfused with 150 mL of compatible

packed red cells and platelets, after which her Hb rose to 8.5 g/dL and platelets to $90 \times 10^9/L$. Following splenectomy, which was performed four months later because of hypersplenism, her Hb rose to 10.2 g/dL and platelets $155 \times 10^9/L$.

The patient was in a steady state for about nine months prior to her death in another hospital during the first week of September 1993. Terminally, she developed paraplegia and was subjected to spinal tap, computed tomography (CT) scan of the head, nerve conduction velocity (NCV) and electromyography (EMG) studies. The CT scan revealed a mild cerebral atrophy with prominent sulci and minimal widening of the third ventricle while the peripheral NCV test confirmed the presence of polyneuropathy.

Case 2. This patient, the younger sister of Case 1, was one year old in February of 1991 when she presented with vomiting, diarrhea, fever, cough, and difficulty breathing. She was pale, lethargic and in respiratory distress with chest retraction. There was hypopigmentation of her skin and hair as well as retinal depigmentation while her spleen and liver were 5 cm and 4 cm below the respective costal margins. She also had small palpable lymph nodes in the cervical and axillary regions. Her peripheral blood smear showed large intracytoplasmic granules in the leukocytes. Bone marrow was not done. Her hematological evaluation revealed a hemoglobin value of 5.7 g/dL, WBC of $17 \times 10^9/L$ and platelets of $143 \times 10^9/L$. Her chest radiograph demonstrated a right upper lobe consolidation and was treated with parenteral ampicillin and oral ascorbic acid for 10 days. She was also transfused with 100 mL of compatible packed red cells with an increase in Hb to 8 g/dL. She was well enough to be discharged home after two weeks of hospitalization.

The patient was readmitted a month later with a history of fever, pallor, lethargy and poor appetite. Her spleen was 9 cm and liver 4 cm enlarged; there were also enlarged lymph nodes and her CBC showed a Hb of 4.3 g/dL, WBC $8.1 \times 10^9/L$ and platelets $12 \times 10^9/L$. She bled profusely from the nose and mouth and was transfused with fresh whole blood but died within hours of admission. Our patients presented with features that were pathognomonic of CHS. These features were composed of oculocutaneous albinism, recurrent chest infections, hepatosplenomegaly, lymphadenopathy and giant intracytoplasmic granules in their leukocytes. While our patients were both females, those of Al-Nasser et al.¹ were all males, thus supporting the autosomal recessive mode of inheritance as reported in the literature.² The parents of our patients are first cousins through their mothers. All four were screened and their peripheral smears showed giant intracytoplasmic granules in their lymphocytes, thus depicting them as carriers even though they did not have clinical features of CHS. Such a simple screening procedure can be used to identify carriers

in a community where there is a high incidence of consanguineous marriages. For those carriers, a gentle marriage counseling can be utilized before a satisfactory prenatal diagnosis becomes available.

The prognosis of this chronic disease is very poor with a high mortality rate in the first decade of life. All the patients so far reported have died within this age period and death has always been due to overwhelming infections or accelerated phase of the syndrome. The accelerated phase of the syndrome in AH was treated with acyclovir, prednisolone and splenectomy with improved clinical and hematological status. This line of management may be recommended, especially for centers where bone marrow transplantation is not practiced.

Adewale A.O. Laditan, FRCP

Department of Pediatrics

Art Yaneza, FRCPATH

Ishwar L. Jeswani, MD

Department of Laboratory Services

King Fahad Hofuf Hospital

Hofuf, Al-Hassa, Saudi Arabia

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