Pure red cell aplasia: presenting at birth

Fatelrahman Ahmed, MRCP (UK), Ranjan K. Pejaver, MRCP (I)
Irfan M. Qureshi, MRCPATH, Maurice Wooldridge, FRCP

Abstract An infant is described who had an Apgar of zero and who was successfully resuscitated. The hemoglobin was 1.5 gm. Pure red cell aplasia was diagnosed. The infant died 16 hours after birth. Pure red cell aplasia presenting in the newborn period is rare. It should be considered while investigating anemia in the neonate.

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Pure red cell aplasia (PRCA) is an uncommon cause of anemia in childhood. The usual presentation is pallor in the first year of life. Red cell precursors are at a low level in the bone marrow. Diamond and Blackfan were one of the first to describe the condition.1

A neonate is presented that was born technically dead. Successful resuscitation was accomplished. A profound anemia was recorded. Despite intensive therapy the infant died 16 hours after birth. A bone marrow examination confirmed the diagnosis of PRCA.

Case Report Male infant A was born with a zero Apgar following an emergency cesarean section for fetal distress. Birth weight was 3.26 kg. Meconium stained liquor and a “flat” cardiocotachogram recording had been noted 15 minutes before delivery. This was the mother's fifth pregnancy. She had taken no medications. No infections or febrile episodes had been recorded. Four other children in the family were normal. The parents were first cousins.

After delivery of the baby, resuscitation procedures were initiated. The infant was intubated, and ventilated with oxygen. External cardiac massage was applied and 0.5 mg of 1/1000 adrenaline was instilled down the endotracheal tube. After two minutes a pulse of over 100 was recorded. No spontaneous respirations were present. The infant remained pale, failing to “pink up”. An umbilical arterial catheter was inserted. The blood was noted to be “thin” resembling colored water. The hemoglobin was 1.5 gm/L. Blood gases showed a pH of 6.5, PCO2 39.1 mmHg, PO2 77.8 mmHg, HCO3 2.9 mmol/L, base deficit 29.9. Sodium bicarbonate was given over the next hour, to a total of 50 ml (4.2% solution). The pH with this therapy could only be raised to 6.9 maximum during the infant’s life span despite further Na HCO3. During the initial resuscitation, blood pressure was maintained at 80/50. Feeble respirations were recorded. Eighty cubic centimeters of whole blood was given over 20 minutes. Stability was established. The infant appeared to have a strengthening respiratory drive.

The external appearances of the infant were normal except for a mild edema. Despite the initial success of resuscitation the infant sustained a series of cardiac arrests. The last occurred 16 hours after delivery. No further resuscitation was possible. The Kleihauer test done on the mother was negative. Blood group testing of the mother and baby did not reveal any evidence of incompatibility. Coombe's test was negative.

A bone marrow examination prior to death, aspirated from the right tibial tuberosity, showed very little erythropoiesis. No orderly maturation through the early red-cell precursors was noted. Most exhibited dyserythropoietic features. Other components of bone marrow were normal for age. The reticulocyte count in the initial film was 0.01%, red cells scanty (0.4 x 10^12/L) and macrocytic. The white cell count was 6.4 x 10^9/L with no abnormal cells seen. Platelets were 76 x 10^12/L.

From the Department of Pediatrics (Ahmed, Pejaver, Wooldridge) and Department of Pathology and Laboratory Medicine, North West Armed Forces Hospital, Tabuk, Saudi Arabia (Qureshi).

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Address correspondence and reprint request to: Dr. R. Kumar Pejaver, Department of Pediatrics, North West Armed Forces Hospital, P.O. Box 100, Tabuk, Saudi Arabia. Fax: (966-4) 422 2324.
Discussion

The diagnostic criteria for PRCA have been defined as 1) normochromic often macrocytic often macrocytic anemia with reticulocytoopenia, 2) a bone marrow with normal cellularity and a deficiency of red-cell precursors, 3) white cells that are normal with a possible decrease in count, 4) platelet counts that are normal. The infant exhibited these criteria, particularly the first three. The diagnosis was made within these parameters.

The etiology of PRCA is obscure. An inherited cause is suggested, although the inheritance is uncertain. Congenital anomalies may be associated with PRCA in 30% of individuals. The commonest is short stature. No anomalies were noted externally in the infant described. Growth was appropriate for a term infant in the given population.

In 10% of children with PRCA there were problems in the pregnancy such as toxemia, hemorrhage and febrile illnesses. Mothers of affected children have further reported increased incidences of miscarriages and stillbirths. The infant described was technically a stillbirth. Bock et al describe a mother with PRCA who had given birth to two infants. Both died from fetal hydrops and anemia. A third pregnancy was successfully treated in utero with three fetal intravascular transfusions. Delivery was successful. The infant was normal at six months old.

Cats viremic with feline leukemia virus subgroups C develop PRCA characterized by the loss of detectable late erythroid progenitors in marrow culture. A viral etiology of PRCA in humans must be considered. Human parvovirus B19 is a strong candidate. Maternal infections produce fetal anemia, aplastic crisis and death. Human parvovirus does not always produce PRCA during pregnancy, although its affinity for red cell precursors is well established. In the case described, the problem appeared to be in the early red cell precursors and not the loss of late erythroid progenitors, as seen in parvovirus infections. However, facilities to perform serological diagnostic tests for parvovirus were not available. No hemolytic anemia or process was observed in the indexed case. The PRCA in infants recorded elsewhere was associated with hydrops fetalis. In the infant described, edema was not a prominent feature. It has not been possible to reproduce hydrops fetalis by simple anemia in the animal model. Other factors must be involved.

References

نوصف حالة هذا الطفل بأنه قد ولد ومجموع علاماته الحيوية صغر، وتم إنقاذه بنجاح. كانت نسبة الهيموجلوبين في الدم 5.1 جم. وتم تشخيص وجود خلايا دم حمراء لانسيجية. توفي الطفل بعد 16 ساعة من الولادة.

إن وجود خلايا دم حمراء لانسيجية في حديثي الولادة بعد حالة نادرة، ويجب أخذ ذلك في الاعتبار عند تقصي حالات فقر الدم (الأنيميا) في حديثي الولادة.

الكلمات الرئيسية: خلايا دم حمراء لانسيجية. حديثي الولادة.