

Case Presentation of Ellis-van Creveld Syndrome and the Dilemmas and Implications of Prenatal Diagnosis and Management of a Congenitally Malformed Baby

Malika Haque, MD, FAAP
Columbus, Ohio

DOI: <http://dx.doi.org/10.5915/30-3-9828>

Abstract

Characteristic features of Ellis-van Creveld syndrome, the dilemmas in its prenatal diagnosis, and the implications in the management of a congenitally malformed baby born to Muslim parents who were second cousins, are described. Following an ultrasound during the third trimester, the parents were told that the baby had a severe congenital malformation called short rib polydactyly syndrome and would die soon after birth. However, the baby continued to live showing improvement without any significant medical interventions, and the diagnosis of Ellis-van Creveld syndrome was considered in the first week of life when first examined by the primary care physician. A diagnostic workup later confirmed the diagnosis. In the fourth week of life, deterioration set in. Despite all the efforts and help from modern medical technology, the baby did not survive and died around nine weeks of age. Both parents and the physicians involved in the care of this infant were faced with difficult dilemmas and decisions in the management of this congenitally malformed baby.

Key words: Ellis-van Creveld syndrome, autosomal recessive inheritance, medical ethics, Islamic jurisprudence.

Ellis-van Creveld syndrome, also known as chondroectodermal dysplasia, was first described by Ellis and van Creveld in 1940.¹ By 1964, about 40 cases were reported. McKusick et al reported 52 cases from

the Amish populations, wherein intermarriages among relatives are quite common.² At present, more than 200 cases of this syndrome have been reported.

Ellis-van Creveld syndrome clinically resembles Jeune thoracic dystrophy or asphyxiating thoracic dystrophy, which are characterized by a small thorax, short limbs, and hypoplastic iliac wings. It also clinically resembles short rib polydactyly syndrome, Type I and Type II. All these syndromes are autosomal recessive in origin and have a poor prognosis, with infants dying soon after birth, except for Ellis-van Creveld syndrome, wherein about half the children live to adulthood.

Case presentation of Ellis-van Creveld syndrome and the medical and ethical dilemmas incurred in the management of a baby boy born to Muslim parents who were second cousins, form the content of this effort.

*From the Department of Pediatrics
and International Health
College of Medicine
The Ohio State University
Columbus Children's Hospital
Columbus, Ohio*

*Reprint Requests: Malika Haque, MD, FAAP
Columbus Children's Hospital
700 Children's Drive
Columbus, OH 43205*

Case Report

A Muslim mother, 18 years old, gravida I, and father, 32 years old, were told by their obstetrician during the seventh month of pregnancy and following an ultrasound examination that their baby had a severe congenital malformation called short rib polydactyly syndrome, which would result in the baby's death soon after birth. Due to short ribs and the associated pulmonary hypoplasia, death occurs in all babies a few hours after birth. No babies with the above diagnosis have survived the neonatal period. Due to poor prognosis of the above syndrome, the obstetrician discussed, "do not resuscitate" (DNR) procedures with the parents who consented to it even though it was a difficult decision for them to agree upon.

On further discussion, it was determined that the family history was very significant for there were many marriages between first and second cousins in the total family structure. On both sides, there was a history of individuals born with short stature and polydactyly, and there was a report of a cousin on the father's side who died soon after birth. There was a history of adolescent cousins on each side with short stature, polydactyly, who were living and reported to be in good health. The parents were well aware of the above characteristics of the syndrome and prepared themselves for the inevitable fact that the baby had a poor prognosis and would not live long.

A baby boy weighing 2888 grams with an Apgar score of one at one minute, five at five minutes, and eight at ten minutes, was born following a spontaneous vaginal delivery. Do not resuscitate procedures were maintained in the delivery room. Much to the surprise of the attending physician, the obstetrician, the neonatologist, and the house staff surrounding the baby, the baby did not take his last breath in the delivery room, but continued to live. The color improved after his first breath, respirations were rapid, about 100/minute. The baby was transferred to the newborn nursery with the assumption that he would die in the next few hours, but he continued to live. Do not resuscitate procedures were still being followed in the newborn nursery. The baby did manifest the characteristic features that are common in short rib polydactyly syndrome, such as having six fingers on each hand and a small thorax with short ribs. After three days in the newborn nursery and when the baby did not die despite DNR procedures, the neonatologist was convinced that the baby did not have short rib polydactyly syndrome and considered a diagnostic workup to explore possibilities of other syndromes.

The parents of the baby did not want to subject the baby to diagnostic procedures and prolonged hospitalization. They wanted to take the baby home and enjoy him in the comfort of their home. The baby was discharged on the third day of life, and the parents contacted the author immediately after the discharge, asking her to be the primary care pediatrician for their baby. The author saw the baby on the fifth day at the Children's Hospital satellite health cen-

ter, where primary care is provided. Physical examination indicated his weight was 6 lbs, 5-6 ozs; length was 17 inches; head circumference was 13 3/4 inches; and chest circumference was 11 inches. He had a characteristic short stature with a small narrow thorax, six short fingers on each hand with nail hypoplasia of fingers as well as toes, a short upper lip with single ridge on philtrum, and a lip that was bound to the gum margin by the frenulum. Also, the baby was voiceless. He was seen again on the eighth day and showed continued improvement and weight gain. Respirations still remained high, between 80-100/minute. The author considered the diagnosis of Ellis-van Creveld syndrome following the initial evaluation and discussed this with the hospital geneticist who agreed to do further diagnostic testing if the baby's condition continued to remain stable by two weeks of age.

At two weeks of age, the baby was admitted to Columbus Children's Hospital for a diagnostic workup, where the geneticist confirmed Ellis-van Creveld syndrome. Several subspecialists, including the geneticist, a pulmonologist, cardiologist, and an ENT, evaluated this child.

Cardiac Evaluation

An EKG with a right shift indicated right ventricular hypertrophy. Echo showed the heart shifted to the right, along with small atrial septal defect and patent ductus arteriosus. Doppler findings were consistent with pulmonary hypertension.

Renal Evaluation

Ultrasound examination of the kidneys showed that they were small with mild dilatation of the renal pelves. Blood urea nitrogen (BUN) and serum creatinine levels were within normal limits.

Radiologic Findings

An x-ray of the long bones showed the right femur to be slightly shortened. X-ray of the pelvis showed a trident configuration, consistent with Ellis-van Creveld syndrome. The humerus and hands were also short, with flaring of metaphyses. An x-ray of the hands showed a postaxial polydactyly with an accessory digit adjacent to the fifth finger. The digits appeared to have very short and broad metacarpals and proximal phalanges. The impression was postaxial polydactyly. These radiologic changes have been described earlier in Ellis-van Creveld syndrome.^{3,4}

Ear, Nose, and Throat Examination

An ENT examination using a nasopharyngeal scope indicated normal vocal cord mobility, normal epiglottis, absence of laryngomalacia, and the presence of an anterior laryngeal web that contributed to the baby's lack of voice when he cried.

Pulmonologist

The pulmonologist confirmed pulmonary hypoplasia:

and a small thorax, small lung tissue, and oxygen saturation ranged between 80 and 90 torr.

Following the initial in-depth evaluation at two weeks of age, the baby was discharged with the recommendation to use supplemental oxygen whenever indicated.

After discharge, during the third week, the baby started experiencing feeding problems, and had to be hospitalized to rule out sepsis as the cause of poor feeding. During this second hospitalization, the baby was found to have a gastroesophageal reflux, and there was no evidence of infection. The baby was sent home on Cisapride and Zantac after three days of hospitalization. The next three weeks of the baby's life were stormy, requiring four hospitalizations, due to poor feedings, and respiratory distress. Two of the last four hospitalizations were in the pediatric intensive care unit. He went on a respirator during the fifth hospitalization for about 48 hours, stabilized, and was discharged to be readmitted again within a week with increasing respiratory problems. During the last hospitalization, his condition continued to deteriorate, and he developed *E. coli* urinary tract infections, staph, and strep tracheitis. He was on the respirator for nine days. Though the infections were totally controlled by appropriate antibiotics, his condition failed to improve. Several care conferences were held with the parents, primary care pediatrician (the author), and other subspecialists to keep the family informed about medical interventions and the step-by-step outcome, and also to prepare them for the inevitable outcome if all the appropriate interventions would fail.

After nine days at the pediatric intensive care unit with appropriate interventions, respirator therapy failed to improve the baby's condition due to pulmonary hypoplasia and a small thorax. It was then mutually decided by the parents and attending physicians to take him off the respirator. He died shortly after he was taken off the respirator after having lived for about nine weeks.

Discussion

Ellis-van Creveld syndrome closely resembles short rib polydactyly syndrome. Both syndromes consists of similar chest pathology, i.e. narrow thorax, short horizontal ribs. In addition, both have polydactyly of the hands. An accurate diagnosis is extremely difficult in the prenatal period due to the close similarity of the syndromes. The parents agreed to go along with the obstetrician's recommendation of not resuscitating when they were told this baby would die soon after birth. When the baby survived beyond the neonatal period and into the second week without any medical intervention and continued to grow despite mild tachypnea, it was necessary to do an in-depth evaluation in order to establish an accurate diagnosis, and any necessary interventions as the diagnosis of Ellis-van Creveld syndrome seemed to be a more likely diagnosis. It carries a better prognosis as 50% of these children grow up to be adults with normal intelligence. Both physicians and parents

agreed to do everything to help the baby grow and achieve his normal potential; therefore, all necessary interventions and medical management was provided. When, despite all the optimum care that modern science and technology could provide, the baby's condition deteriorated, physicians and parents agreed to take the baby off the respirator. In this particular case, the baby's narrow thorax and lung hypoplasia could not prolong life further.

The parents and physicians understood the limits of medical intervention. This was also in agreement with the religious ethics of the Muslim parents, which do not approve of heroic measures to artificially prolong life of a terminally ill patient.⁵ Genetic counseling also was provided to the parents regarding future pregnancies. They were told that there was a 25% chance with each pregnancy for them to have a child with Ellis-van Creveld syndrome. They were also told that 50% of their children, though phenotypically appearing normal, would genotypically be carriers of autosomal recessive trait of Ellis-van Creveld syndrome. They were also told that 25% of their children would be genotypically normal.⁶ The parents accepted the facts that were conveyed to them on genetic counseling.

In view of the fact that the parents, as well as the primary care physician are Muslims, genetic counseling, in addition to being factually presented by the Muslim physician to the Muslim parents, had to be complemented by Islamic beliefs. Though there is no prohibition of prenatal diagnosis, some could argue its redundancy because even if the fetus is found to be suffering from a severe abnormality, abortion will not be an option, unless it is to save the mother's life. However, others may consider abortion permissible in such cases, especially in early pregnancy. This is truly a modern dilemma and requires further study by Islamic scholars and physicians with expertise in this field. The parents with faith and trust in Allāh (SWT) may decide to decline prenatal diagnosis and pray that their children will not be affected.

References

1. Ellis RWB, van Creveld S: A syndrome characterized by ectodermal dysplasia, polydactyly -chondro-dysplasia congenital morbus cordis. Report of three cases. *Arch Dis Child* 1940;15:65-70.
2. McKusick VA, et al: Dwarfism in the Amish. The "Ellis-van Creveld syndrome." *Bull Hopkins Hosp* 1964;115:306-8.
3. Taylor GA, et al: Polycarpal and other abnormalities of the wrist in chondroectodermal dysplasia; The "Ellis-van Creveld syndrome." *Radiology* 1984;151:393.
4. Qureshi F, et al: Skeletal histopathology in fetus with chondroectodermal dysplasia ("Ellis-van Creveld syndrome"). *Am J Med Genet* 1993;45:471-2.
5. Athar S: Islamic Perspectives in Medicine. American Trust Publications 1993:189.
6. Behrman RE, et al.: "Nelson Text Book of Pediatrics," 14th edition. WB Saunders Company 1992:272-3.