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A severe case of Beare-Stevenson syndrome and associated congenital deformities

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SUMMARY. **A severe case of congenital anomalies is described, with several characteristics of Beare-Stevenson syndrome, such as cutis gyrata, acanthosis nigricans, craniofacial anomalies, ear defects, enlarged umbilical stump and anogenital anomalies. He does not have craniosynostosis or clover leaf skull, which has also been described in this syndrome. This patient also shows hands and feet anomalies, absence of skin adnexa in several locations and dental anomalies, which could suggest an associated ectodermal dysplasia.**

Case report

A male of mixed race (white, Indian and black) was born by natural delivery after a nine month pregnancy, in a small town in the north of Brazil. The mother was 23 years old in good health, with no physical defects, and the father could not be located. There were reports from the local hospital that the mother tried to abort after the second month of pregnancy by ingesting plant infusions of unknown composition. The child was abandoned at the hospital 3 days after the delivery and the mother has not been located since then. It was her first child and no family history was obtained. At birth, the local doctor noted, besides the obvious congenital defects, raw areas in the forehead, abdomen, hands, wrists, ankles and feet, in a very symmetrical position (Fig. 1). The child found breathing and feeding difficult and required special nursing care. The raw areas healed by secondary intention and he was kept in good condition up to one year when he was sent to our service.

Physical examination

The internal organs did not show any abnormalities. The skull was normal. There was ocular hypertelorism. The eyes were exposed, caused by a skin retraction involving the eyebrows, upper lids and skin of the forehead. The lower lids were displaced laterally, with complete ectropion. The cornea was protected by frequent eye movements and during sleep by a strong Bell's phenomenon. There was a fissure extending from the medial canthus to the alveolar ridge. The palate was closed. This fissure seemed to be part of the lacrimal apparatus. The nose was absent, with soft

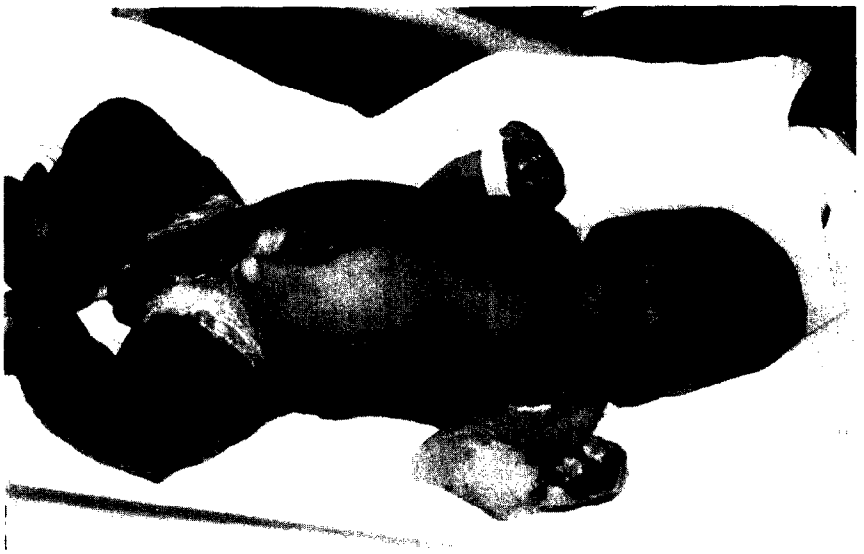
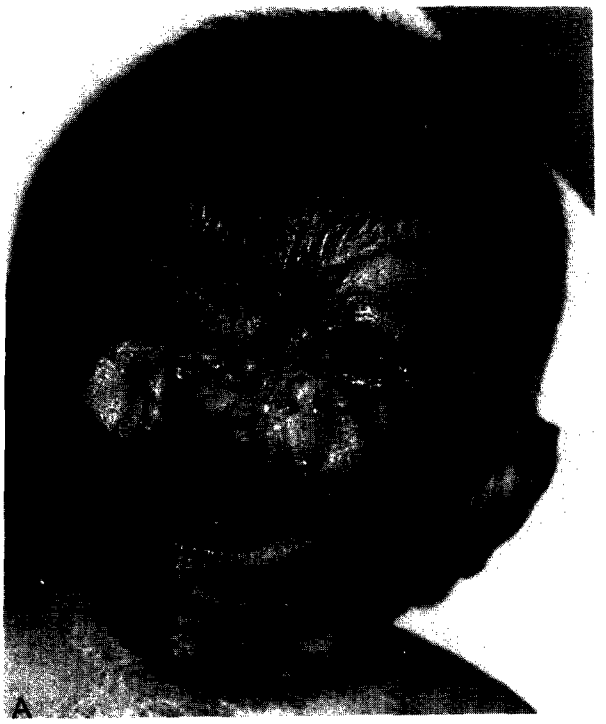


Fig. 1

Figure 1—Copy of a photograph taken at the hospital where the child was born. Note the raw areas on the forehead, abdomen, wrists and ankles, prominent umbilical stump, and other congenital defects. The eyebrows were not retracted and the defects of the hands not so severe.

Fig. 2

Figure 2—Patient one year old. Full body view.



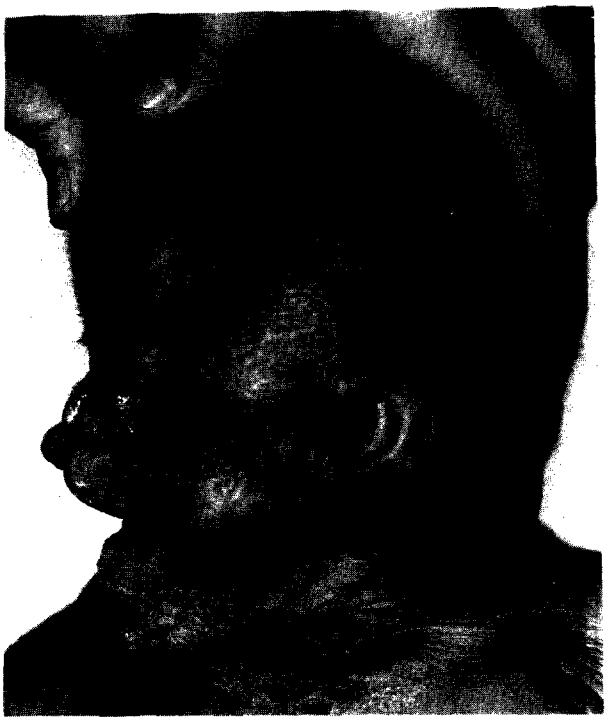


Fig. 3

Figure 3—(A, B). Front and lateral view of the face at one year. Notice that the eyebrows and upper eyelids are now much higher than in Figure 1. The other defects are described in the text.

tissues and hairs at the upper part. The lower part showed structures similar to embryological pits but the nostrils were unperforated. Below this area there was a prolabium overlying a closed maxilla with a small median cleft. The lower lip was closed but there was a triangular area of tissue in the midline, extending down to the chin. The ears were malformed, with open auditory meatus. Hearing seemed normal. Cutis gyrata was present on the forehead, cheeks and part of the scalp, which also showed superficial scars and thin hair. At the parietotemporal region bilaterally, there was a skin crease resembling the primitive olfactory placode (Figs 2, 3). The posterior skin of the neck was normal, showing only small pits over the cervical vertebra. The abdomen had an enlarged umbilical stump, surrounded by a pigmented area, extending from the chest to the pubis (Fig. 4). The scrotum was bifid and the raphe extended to the anal area, which was in a more anterior position. The skin of the penis had a circumcised appearance, exposing the glans (Fig. 5). Cutis gyrata was present on both legs, with deep creases on the thighs and buttocks (Fig. 6). Acanthosis nigricans was present on both extremities. The palms and soles had furrows and a partial syndactyly. The fingers were in flexion with malposition of the thumbs, which seemed to be caused by scar contraction. The distal phalanges were free and the nails hypoplastic with a pointed shape. The feet showed a similar defect (Figs 7, 8). A radiograph of the face showed an anterior bony obstruction of the nasal cavities, which were open posteriorly. A three dimensional CT (Fig. 9) and magnetic resonance imaging showed this abnormal bony plate in the nasal region and maxilla, closing the nasal cavities anteriorly, and adenoidal type of tissue partially obstructing these cavities posteriorly. The radiographs of the arms, legs, hands and feet did not show any abnormalities in the long bones. The phalanges were normal in size, but in

Fig. 4

Fig. 5

Figure 4—The umbilical region. Enlarged umbilical stump, acanthosis nigricans. Scars in the midline and lateral areas after healing of the raw tissue found at birth. **Figure 5**—The skin of the penis and scrotum.

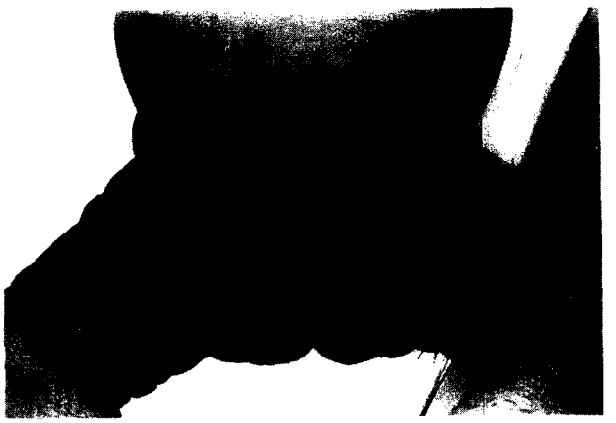


Fig. 6

Fig. 7

Fig. 8

Figure 6—The skin of the buttocks and thighs (cutis gyrata and acanthosis nigricans). **Figure 7**—The hands. The displacement of the thumbs, as compared with the newborn pictures, seemed to be aggravated by scar contraction. **Figure 8**—The feet, showing acanthosis nigricans and furrows at the soles, skin appendages with a small bone inside and displacement of the hallux. Both feet were absolutely symmetrical.

a distorted position and at both ankles there was a small bone not connected to the tibia. Biopsies of the skin were done at several locations of the head, trunk and extremities. The palms and soles revealed acanthosis nigricans, also present at the legs, abdomen and chest. The corrugated skin of the face, umbilical region, inner part of the thighs and the buttocks showed a picture compatible with cutis gyrata, besides some degree of acanthosis, hyperkeratosis and an absence of sweat and sebaceous glands. The abnormal tissues in the lower lip showed epidermis with acanthosis and parakeratosis, disorganised striated muscle fibres, vascular hyperplasia and general fibrosis. The scalp showed normal structures, including hair follicles; in the temporal region the follicles and sebaceous glands were atrophic. The laboratory examinations were normal. A karyotype showed the following: 46XY,t(7;18)(q35;q21), *i.e.* a balanced translocation between the long arms of chromosomes 7 and 18. We were unable to get blood from the mother.

Progress

This patient was sent to our service when he was one year old. Since then, he has been under observation for 6 years. Initial surgery to open the nostrils was done at the age of 14 months to help breathing and feeding. Because of the increased surgical risk, partial closure of the upper lip, correction of the lower eyelids and removal of abnormal tissue of the lower lip was delayed until the child was 5, as well as correction of the position of the thumbs. The lower teeth were present

Fig. 9

Fig. 10

Figure 9—CT of the face showing the bony plate anteriorly in place of the nasal bones and part of the maxilla. One nostril had been opened surgically. **Figure 10**—Patient 5 years old showing the eruption of the upper teeth. Both nostrils were open, with tubes in place to prevent stenosis.

after the second year of age, but the upper teeth started to erupt only after the fourth year, beginning with the canines (Fig. 10) and later the lateral incisors. His speech is very poor probably because of the still open upper lip and a high palate. However, neurological development, growth, muscular coordination and walking seem normal. Surgical correction of the remaining defects has been planned and will be reported later.

Discussion

This case is very dramatic because of the serious defects found and the association of different congenital anomalies. After research in the literature it was concluded that this is a severe case of Beare-Stevenson syndrome.[1,2] In a personal discussion with R. J. Gorlin (1992), we came to the conclusion that it was the most complete case of this syndrome described in the literature. However, the histological findings of absence of sweat and sebaceous glands in several locations, teeth abnormalities, nail defects and other malformations in the face, could suggest some form of associated ectodermal dysplasia.[3]

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