A Variant of the Wiedemann-Beckwith Syndrome

W. Wöckel, K. Scheibner, and A. Lageman

Pediatric Pathology Division (Head: Dr. W. Wöckel, MD), Department of General Pathology and Pathological Anatomy (Director: Prof. D. Schreiber, MD), Medical Academy Erfurt, Nordhäuser Str. 74, DDR-5060 Erfurt, German Democratic Republic

Abstract. The following combination of findings were established by histological examination of biopsy or autopsy material in an 11-day-old male baby: congenital mesoblastic nephroma, nodular renal blastema, dysgenetic pancreatic cyst, "cytomegaly" of the adrenal cortex, hyperplasia and hypertrophy of the islets of Langerhans, hypoplasia of the thymus and the lymphatic tissue. Since some of these changes constitute the characteristic histological findings in the Wiedemann-Beckwith syndrome and the remainder can be readily reconciled with it, the assignment of this case to this syndrome is discussed. Although the three cardinal symptoms are lacking, the combination of findings are interpreted as a variant of the Wiedemann-Beckwith syndrome.

Key words: EMG syndrome - Exomphalos-macroglossia-gigantism syndrome - Fetal adrenocortical cytomegaly - Congenital mesoblastic nephroma - Nodular renal blastema - Variant of Wiedemann-Beckwith syndrome - Wiedemann-Beckwith syndrome.

In 1964, Wiedemann [41] as well as Beckwith and co-workers [4] independently described a combination of findings which are now known as the Wiedemann-Beckwith syndrome [24]. This syndrome is characterized by the symptom triad of exomphalos, macroglossia and gigantism and is therefore also referred to as the EMG syndrome [12, 16, 24, 30, 36, 43]. In addition, there are further external and internal malformations as well as other abnormalities (reviews at [12, 16, 34]).

Case Report

Clinical Data

A laparotomy had been performed during pregnancy on the mother of the child in another hospital because of ileus: a "dermoid cyst" of the ovary had been removed. After the operation, there was premature rupture of the fetal membranes and birth. The male baby born before term (duration of pregnancy: 35 weeks) had a birth weight of 2850 g and a length of 50 cm. A tumor of the left kidney was diagnosed on the first day of life. Since there was severe distress, left nephrectomy could only be performed on the tenth day of life. At the same time, a pancreatic cyst was removed. Postoperatively, there was respiratory arrest and cardiac arrhythmia. In addition, there were pulmonary hemorrhages and bronchopneumonia. The baby died two days after the operation.

Morphological Findings (KS Nr. 82/77)

The post mortem findings were as follows: nephrectomy for a left sided congenital mesoblastic nephroma 5 x 5 cm in size (Figs. 1 and 2) as well as removal of a dysgenetic pancreatic cyst (A-No. 1649-50/77). Nodular renal blastema (Fig. 3) of the right kidney, as well as of the remaining tissue of the left kidney. Postoperative circulatory failure with focal necrosis of the liver and spleen, as well as hemorrhagic necrotizing enteropathy with terminal perforation of the jejunum (intestinal contents in the abdomen, but no peritonitis). Small subependymal hemorrhage in the right lateral ventricle. Focal intra-alveolar pulmonary hemorrhages as well as patchy bronchopneumonia. "Cytomegaly" of the fetal zone of the adrenal cortex (Fig. 4). Hyperplasia and hypertrophy of the islets of Langerhans (Fig. 5) (normal blood sugar values according to clinical data although blood sugar was only determined once). Hypoplasia of the thymus and lymphatic tissue.

1 We thank Professor H. Patzer, MD, Director of the Department of Pediatrics and Professor E. Gottschalk, MD, Head of the Pediatric Surgery Division, Department of Surgery (Director: Prof. W. Usbeck, MD), Academy of Medicine in Erfurt, for providing us with the clinical results.
Discussion

Of the morphological findings, the changes in the adrenals, the pancreatic islets system and the remaining kidney can be regarded as components of the Wiedemann-Beckwith syndrome [12, 17, 39]. The question thus arises as to whether our case should be assigned to this syndrome. "Cytomegaly" of the fetal zone of the adrenal cortex, which has nothing to do with cytomegalovirus infection [22] and the significance of which is not clear [11, 27] occurs regularly in primary congenital and in primary cytotoxic adrenocortical hypoplasia [6]. However, this can be excluded in our case. It has been described repeatedly in Wiedemann-Beckwith syndrome [1, 4, 6, 16, 19, 26, 29, 31, 32, 33, 34, 36, 39], often combined with a hyperplasia of the islets of Langerhans [1, 4, 16, 29, 31, 33, 34, 36, 39]. A few other authors have also observed islet cell hyperplasia in this syndrome [10, 35] as well as a nesidioblastosis [10, 21, 31]. These changes in the islets, as well as metabolic studies, indicate presence of hyperinsulinism [12] and explain the abnormal glucose tolerance test, which is occasion-

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1 The Figs. 1–3 derived from the study by Wöckel et al. [47]