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# *Brachmann's syndrome (Cornelia de Lange's syndrome)*

Brachmann's (Cornelia de Lange's) syndrome is a rare defect characterized by numerous abnormalities and various degree mental retardation. It occurs once per 50–100 thousand births (1, 5).

The etiology has not been explained explicitly yet. The possible causes include: dominant autosomal heredity; it might be related to the duplication of the third chromosome long arm (7). The complexity of the defect may be due to embriopathy of unknown origin developing in the early period of fetal life (3).

Typical characteristic features include: low birth body weight, short size (dwarfism), delayed physical development, facio-cranial dismorphia, microcephaly. Remarkable are facial features, so--called "clown's face" with low forehead, abundant hair (hirsutism), thick eyebrows grown together, long curly eyelashes, antimongoidal wide lid slits (hypertelorism), small flat nose, prominent philtrum, thin lips with lowered corners and low set auricles (2, 3, 7, 8, 10).

Other characteristics are: muscular hypotonia, contractions and limited range of movement in the elbows and hips, vertebral defects, shortened long bones, polidactyly (extra fingers), oligodactyly (lack of fingers), syndactyly (fingers grown together), clinodactyly (finger contractions with adductant thumbs), extra ribs, completely missing arms (2, 3, 9, 10).

In addition to that, the syndrome includes defects of the heart, digestive system, middle ear and diaphragmatic hernia. There are also other problems like complicated feeding, irritability, low husky cry and susceptibility to recurrent infections of the airways, middle ear and skin due to insufficiency of the immune system (6, 7, 10). The literature also reports microgenia, "gothic palate" and cleft palate (2, 4, 9).

Mortality has not been determined precisely, though a considerable percentage do not survive adulthood due to both respiratory infections and cancer that develops as a result of impaired immunity. Besides, many early deaths occur as a result of anomalies within the brain (10).

## CASE STUDY

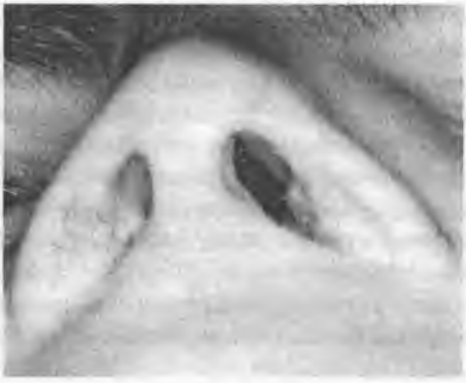
The patient: a boy (G.T.) born in February 1989 was first brought to The Chair and Department of Paediatric Dentistry, Medical University of Lublin in 2004. When seen, the patient was 15 years old. At the age of 6, in February 1995, he was consulted in the Paediatric Clinic, Medical University of Lublin. The examination found physical development considerably delayed in relation to his age group: at that time the boy's height was at the level of a 2-year 3-month-old baby, his body weight of an 8-month old, head circumference of a 5-month old, chest circumference of a 5-year old, chest

width of a 3-year old baby; only chest depth was ahead the real age, i.e. of an 8-year old child. On admission the patient was 1.25 m tall, body weight 18 kg, of disharmonic built, with muscular atrophy, skinny, not able to walk by himself (Phot. 1).

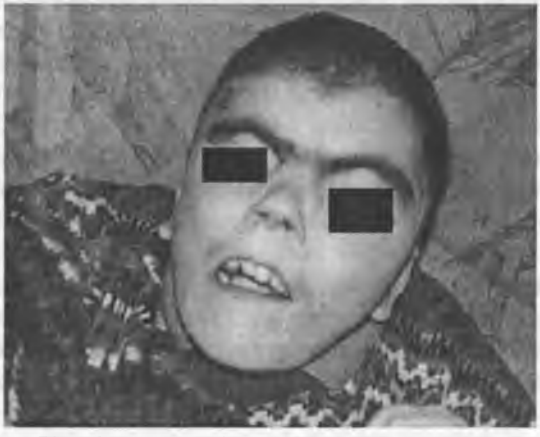
Phot. 1. General look at the patient

Family history revealed that his mother reported uneventful course of pregnancy and pre-term spontaneous delivery. The examination carried out after birth found numerous congenital birth defects of the bones, heart defect, capistration, bilateral inguinal hernias. US scan of the head taken in May 1989 detected normal unwidened ventricular system of the brain, no focal changes in the cerebral tissue except the area of the longitudinal fissure. X-rays of the head found bone loss within the left parietal bone, weak calcification of the cranial covering bones, normal picture of the sella turcica. Cardiological examination revealed atrio-venous shunt at the ventrical level, inclination of the heart electrical axis towards the left, features of hypertrophy of the right ventricle and right branch of his bundle blockage.

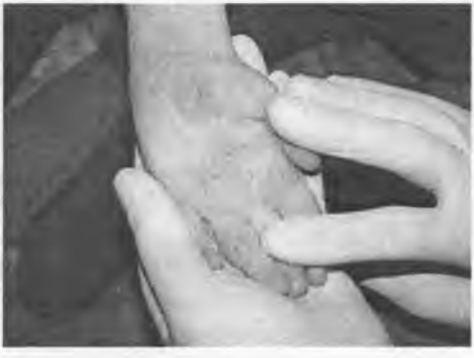
Physical examination revealed pronounced microcephaly with advantage of the facial part over the cranium, small narrow face, wide and grown together eye brows, visibly curved nose (Phot. 2), eyes set wide apart from each other with lowered external corners, shallow set eyeballs, alternating convergent squint. Ophthalmological examination detected bright opacities within the vitreous body in the left and right eye, optic nerve disk more pale and partly opaque in both eyes; anterior chamber of the eye was unremarkable. Other findings were pronounced cheek bones, microstomia (narrow mouth slit), permanently open mouth with dried lips, which was a sign of mouth breathing (Phot. 3). Asymmetric narrow anterio-posteriorly arched chest. X-rays revealed congenital deformations of the ribs, steeply placed upper ribs, extra rib on both sides, large and wide vascular hili, narrow intercostal spaces, limited range of movement in the big joints of the upper limbs with poor muscular mass and small abnormalities of the nail plates and numerous scars on the dorsal and inner side of the hand caused by autoaggression (Phot. 4, 5). Lower limbs presented a limited range of movement, were bent in the big joints, feet directed inwardly, syndactyly of toe 2 and 3 at the proximal phalanges. Skin on the knees thickened, horny and scaling off.



Phot. 2. Curved nose



Phot. 3. Permanently open mouth with dried lips



Phot. 4 and 5. Numerous scars on the dorsal and inner side of the hand caused by autoaggression

Psycho-motoric development of the boy is delayed to a great degree and is global. He can sit by himself but is not able to walk himself. Strong muscular contractions, especially in the big joints of the upper and lower limbs are observed. He attempts to move and climb the furniture. He manipulates objects, though it is limited to single, repeated movements like putting objects into the

mouth, rubbing teeth or furniture with a toy. There is hardly any contact with the child; all attempts at establishing nonverbal contact, even by his mother, are negative. Autoaggression taking the form of beating the head with hands, biting or hitting the head against the furniture is observed. The boy is able to articulate meaningless sounds only. He does not respond to instructions and demonstrates no signs of speech cognition. He requires continuous total care, does not signal physiological needs.

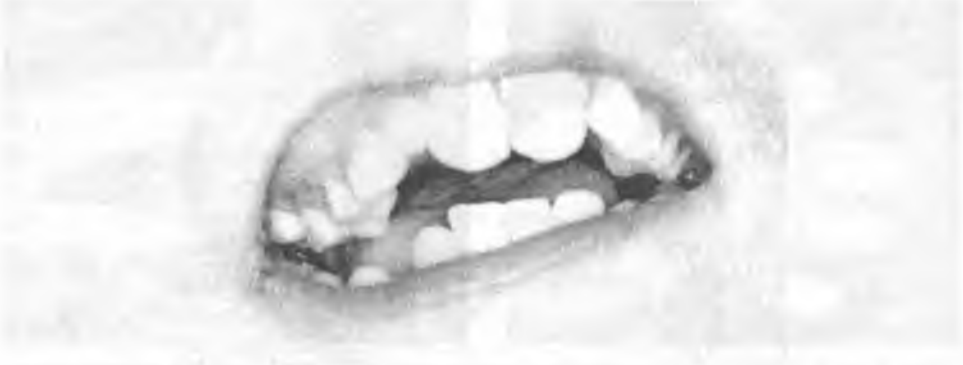
The patient was hospitalized many times at the gastroenterological wards for reflux disease and esophageal hiatus. Endoscopic examination found the esophagus of irregular lumen, stenosed in the middle region, of weakened peristalsis. Mucus lining exhibited signs of inflammations, with numerous erosions bleeding profoundly during the endoscopy. Contrast radiological examination revealed patent alimentary tract with balloon-like dilation at the esophageal hiatus. In October 1995 he was operated on for esophageal hernia, which successfully resulted in the remission of reflux symptoms.

The patient was frequently hospitalized at the pulmonological wards, twice for pneumonia on both sides, in other cases to suck away mucus secretions from the bronchial tree.

Considering child's general condition – Cornelia de Lange's syndrome, with very deep mental retardation, oral treatment was carried out under general anesthesia. It was the first dental treatment in the boy's life.

Intraoral examination, performed under general anesthesia, detected advanced dental caries affecting permanent and primary teeth expressed as DMF and dmf values 10 and 4 respectively. The values were mainly due to the D and d component, i.e. the number of teeth affected by decay. Another finding was advanced bite defect – complete distocclusion with protruding maxillary incisors and narrowed alveolar arch of the mandible, insufficient space for the teeth 33, crowding and rotation of the mandibular incisors, persistent primary teeth: 53, 54, 55, 63, 64, 65, 74, 75, 84, 85. (Phot. 6). In the lower alveolar arch both 7 teeth unerupted. All abnormalities prove disodontiasis. The teeth 12 and 22 had decreased size of their anatomical crowns. Assanation conservative procedures were performed on 12 teeth (7 treated for medium caries and 5 teeth for profound caries), two were qualified for extraction (8).

To sum up, the patient, who is physically handicapped and mentally retarded, remains under continuous dental care with special attention to professional cleaning and fluoride prophylaxis.



Phot. 6. Persistent primary teeth

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## SUMMARY


The authors describe the case of a 15-year-old boy suffering from very rare Brachmann's (Cornelia de Lange's) syndrome, the defect characterized by numerous developmental abnormalities and mental retardation of various degree. The boy remains under specialist care provided by many specialists, including complex dental treatment performed under general anesthesia.


## Zespół Brachmanna (Cornelia de Lange)


Autorzy opisują przypadek 15-letniego chłopca z rzadkim zespołem Brachmanna (zespół Cornelia de Lange), charakteryzującym się wieloma wadami rozwojowymi oraz różnego stopnia opóźnieniem umysłowym. Chłopiec pozostaje pod stałą opieką wielospecjalistyczną, w tym prowadzoną w znieczuleniu ogólnym sanacją jamy ustnej.