

A successful multidisciplinary treatment of a boy with syndromes Down and Cri-du-chat

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Background

Children with Down syndrome (partial trisomy of 21st chromosome) and 5p-(5p minus) (Cri-du-chat syndrome) have a specific phenotype features.

Chart 1. Typical physical characteristics and the most common diseases associated with Down and Cri-du-chat syndromes

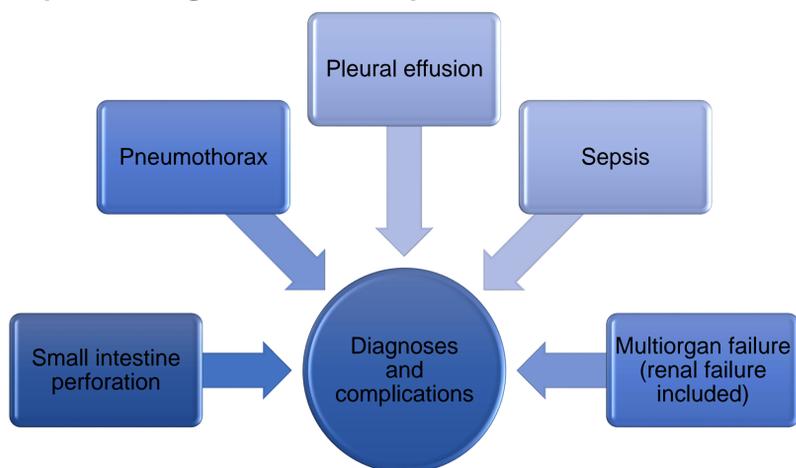
Typical physical characteristics			The most common diseases associated with:	
Down syndrome	Cri-du-chat	Common features of Down and Cri-du-chat syndromes	Down syndrome	Cri-du-chat syndrome
<ul style="list-style-type: none"> ➤ Upward slanting eyes ➤ Flat nasal bridge ➤ Uprturned nose ➤ Epicanthal folds ➤ Small palpebral fissures ➤ Smooth philtrum ➤ Small mouth ➤ Tongue protrusion ➤ Small ears ➤ "Railroad truck" ears ➤ Thin upper lip ➤ Excess skin of the nape of the neck ➤ Wide short hands with short fingers ➤ Separated joints between the bones of the skull ➤ Single crease in the palm of the hand ➤ Decreased muscle tone at birth mental retardation 	<ul style="list-style-type: none"> ➤ Rounded face ➤ Widely spaced eyes ➤ An extra fold of the skin at the inner corners of the eyes (epicanthal folds) ➤ Flattened and widened nasal bridge ➤ Ears that are positioned low on the head ➤ Microcephaly ➤ A short neck ➤ Typical cry ➤ Mental retardation 	<ul style="list-style-type: none"> ➤ An extra fold of the skin at the inner corners of the eyes (epicanthal folds) ➤ Widely spaced eyes ➤ Flattened and widened nasal bridge ➤ Mental retardation 	<ul style="list-style-type: none"> ➤ Ventricular septal defect (VSD) ➤ Hirschsprung disease ➤ Coeliac disease ➤ Hypothyroidism ➤ Diabetes ➤ Alzheimer disease ➤ Leukemia 	<ul style="list-style-type: none"> ➤ Corneal dystrophy ➤ Homocystinuria ➤ Mielodysplastic sindroma ➤ Parkinson disease ➤ Primary carnitine deficiency ➤ Recessive multiple epiphyseal dysplasia ➤ Spinal muscle atrophy

Case report:

Male premature baby with BW 1860 g, length 43 cm, head circumference 31 cm, APGAR score 8/10. The child was presented with common features of Down and Cri-du-chat syndromes (proved later by genetic testing). Because of severe acidosis at birth the baby is treated in Neonatal ICU, additional examination revealed ventricular septal defect (VSD) and periventricular haemorrhage II/III grade.

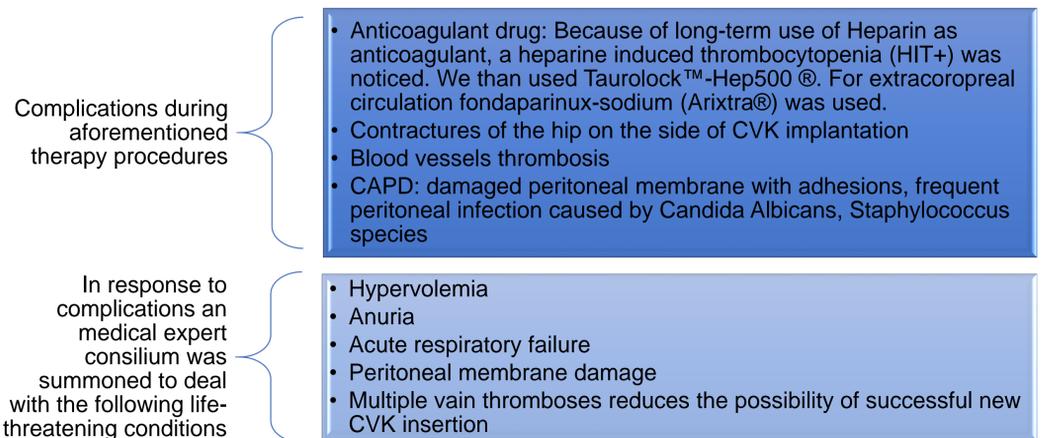
Follow-up and treatment at 1 yr of age. The boy had BW 6,5 kg. After surgical closing of VSD the baby was monitored in pediatric ICU for possible complications. After a few days of routine normal monitoring of vital functions as well as clinical and laboratory testing, a sudden onset of the following complications were noticed intraabdominal bleeding and haemorrhagic shock with need for reanimation and mehanical ventilation.

Graphic 1. Diagnoses and complications



In response to the present complications CRRT was performed via Central venous catheter (CVK)/24 hr/6 month of duration. CVVHD and CVVHDF procedures were performed with: HF 20 set, flow of blood pump: 50 ml/min, flow of dialysate: 300 ml/h, reinfusate flow: 100 ml/h, ultrafiltration rate: 50 ml/h depending of hypervolemia.

Graphic 2. Dialysis complications



Graphic 3. Medical consilium



Conclusions of medical expert consilium

- Once again placement of CVK despite of low possibility of its successful insertion for hemodialysis treatment
- If new CVK insetion will be successful, a close monitoring of blood flow and hemodialysis achievement/performance
- Else, despite its limitations a peritoneal dialysis is the only possible solution left
- Else, sadly but a lethal outcome will be inevitable

Outcome of clinical decisions

- Successful CVK insertion
- Hemodialysis successfully performed
- Improvement of respiratory function with no need for further mechanical ventilation
- Improvement of kidney function with Furosemide therapy
- Graduall cease of hemodialysis
- Full recovery of the child after 1 year of treatment

Final outcome

- The child is independently walking with some additional support (aged 4.5 yr)
- Simple linguistic sentences (6 yr)
- BW 16,5 kg; length 105 cm (6 yr)
- Physical condition according to age
- Normal kidney function
- Multidisciplinary approach for further habilitation continues

Conclusions

