

Table S1. Comparison of present case with cases as reported in de Pontual et al [31].

	Case Presented	Case 1 - de Pontual et al	Case 2 - de Pontual et al
Sex	female	female	female
Gestational age	32 weeks	full term	31 weeks
Birth Weight	1890 g (-0,5 SD)	3100 g	1300 g
Birth Length	43 cm (+/-0 SD)	46 cm	not specified
Head Circumference	31.5 cm (+1SD)	34 cm	28.5 cm
Growth	poor overall growth, deceleration of head growth	deceleration of head growth	poor overall growth, deceleration of head growth
Age at NB diagnosis	4 weeks	3 days	3 weeks
NB presentation	multifocal lobular masses in abdomen and thorax	adrenal NB with pelvic extension, rapid tumor progression	bilateral large heterogenous and calcified adrenal masses
NB histology	undifferentiated in thoracic tumor, ganglioneuroblastoma in abdomen	not specified	infiltrating islands of undifferentiated neuroblasts.
NB genetic profile	non-MNA, numerical only (sample 1&2), other segmental aberrations (sample 3)	non-MNA, non-1p-del (FISH)	non-MNA, 4 copies of MYCN, trisomy 1, trisomy 9, tetrasomy 17 (FISH)
Germline <i>PHOX2B</i> mutation	no	no	no
Germline <i>ALK</i> mutation	F1174I, <i>de novo</i>	F1245V, <i>de novo</i>	F1174V, <i>de novo</i>
Congenital Malformations	diaphragmatic hernia, unilateral inguinal hernia	(high arched palate)	tracheobronchomalacia, bilateral inguinal hernias, patent foramen ovale
Intestinal Findings	hypomotile; no abnormal enteric plexuses	hypomotile; no abnormal enteric plexuses	hypomotile; no abnormal enteric plexuses
Autonomic Instability	frequent episodes of apneas, hypoventilation and desaturation	episodes of apneas and desaturation	daily episodes of desaturation; temperature instability
CCHS workup	polysomnography: apneas independent of epileptic activity; reduced heart rate variability consistent with CCHS	CCHS initially suspected but ruled out due to independence from sleep-awake state and absence of <i>PHOX2B</i> mutation	CCHS initially suspected but ruled out due to independence from sleep-awake state and absence of <i>PHOX2B</i> mutation
Neurologic Symptoms	atrophic muscles, abnormal movements, contractures, possibly deviant eyesight behaviour; neurogenic bladder dysfunction; intact sucking reflex	hypotonia, scarce spontaneous movement, absence of sucking and swallowing reflexes	hypotonia, scarce spontaneous movement, poor sucking, swallowing difficulties; abnormal movements of right arm and leg
EEG findings	epileptiform activity but clinical seizures without corresponding EEG changes	slow activity; no signs of epilepsy	no epileptiform activity
Brain MRI	normal brain MRI including brainstem	abnormal shape of brainstem	abnormal shape of brainstem
Cause of Death	respiratory failure	apnea; no attempt at resuscitation	withdrawal of intensive care
Age at Death	3 months	4.5 months	9 months