What's in a name? – Revisiting the syndromatic appearing child for surgery, post-diagnosis

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Summary Walker-Warburg syndrome is a rare entity. Ethical dilemmas arise in anaesthetizing a premature neonate pre- and post- this diagnosis. We share our concerns of dealing with a syndromatic appearing child and his repeat anaesthetics, post-diagnosis.

Introduction Walker-Warburg syndrome (WWS), also known as HARD (Hydrocephalus, agyria, retinal dysplasia) syndrome, is rare, 1-2:100,000 [1-6]. An autosomal recessive congenital disorder, it is a severe form of the congenital muscular dystrophy, resembling muscle-eye-brain disease (MEB). Diagnosis necessitates four abnormalities: Cobblestone lissencephaly, cerebellar malformation, retinal malformation, and congenital muscular dystrophy [7,8].

Anaesthesia management may be challenging yet is scantly reported [1,2,5.6]. The muscular dystrophy may entail respiratory complications and possibly malignant hyperpyrexia (MH)[9]. Hydrocephalus may cause increased ICP. Maxillofacial malformations (14%) may predispose to a difficult airway [9,10]. The majority die as infants (<1 year old), secondary to the severe defect in brain development. We present a case of repeat general anaesthesia in an infant with WWS.

Report Our patient, MRR, suffered prenatally from a communicating hydrocephalus, corpus callosum agenesis, cerebellar malformation and suspect retinal detachment. Amniocentesis and genetic chip counseling followed without any diagnosis confirmed. MRR was delivered prematurely (35w+5d.) by emergent CS due to bradycardias in a breech fetus. He weighed 2,200g and was macrocephalic (42.8cm). Apgar tests at 5/10min were 6/8. He was admitted to the neonatal intensive care unit (NICU) apathetic and in mild respiratory RDS. His macrocephaly, low set ears and hairline, axial hypotonia and poor suckling defined a "syndromatic appearing child", SAC (Fig. 1) [9,10].

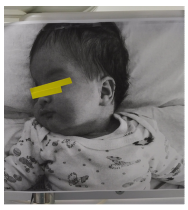

Figure 1

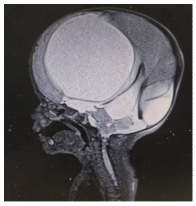

figure 2

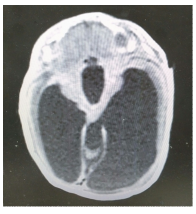

figure 3

He initially underwent two days of non-invasive ventilation. MRI demonstrated bilateral supratentorial severe communicating hydrocephalus, dysplastic optic nerve, cerebellar hypoplasia and z-shaped midbrain (Fig. 2-3). The anesthetic for the MRI was a propofol drip on spontaneous ventilation.

A right sided VP-shunt was scheduled for day three. The patient was induced with sevoflurane, iv fentanyl and atracurium. Intubation was easy. Anesthesia was maintained by volatiles, supplemented with fentanyl and rocuronium bolusi. After an uneventful surgery, the patient was transferred intubated to the NICU. He was kept intubated until 2 days later, failing extubation. He convulsed on day 7 and was put on antiepileptics. The definitive WWS genetic diagnosis arrived on day 29.

We were scheduled to re-anesthetize him on day 53, for a gastrostomy insertion. We were grappling with the new precautions WWS entails. A literature survey regarding second anesthetic exposure in WWS, presented few references citing single exposure, and none multiple exposures [1-3, 5-6, 11-12]. An MEB case series [within ref 1] describes repeated general anesthesia for four patients with volatiles and succinylcholine causing severe increases in CPK levels. We, accordingly, forwent possible MH triggers by utilizing propofol based TIVA supplemented with fentanyl bolusi, despite the uneventful first surgery on volatiles. We also refrained from using paralytics, considering his severe axial hypotonia and the WWS muscular dystrophy. After this second, uneventful surgery, the patient was extubated in the operating room and returned spontaneously breathing and monitored to the NICU.

.On day 118, MRR was sent to community nursing care for PEG feeds, oxygen supplementation and chronic anticonvulsive therapy.

Discussion Walker-Warburg syndrome (WWS) is a rare entity and is a marginalia of this case report. Professional and ethical dilemmas arise in anesthetizing a premature neonate pre- and post- his disorder's diagnosis. We surface the original concerns of dealing with an SAC [4] and the problematics of repeat anesthetics to a recently defined pathology. Our patient, MRR, had been known prenatally to suffer from brain anomaly. The definitive WWS genetic diagnosis arrived on day 29. Muscle-eye-brain (MEB) disease shares many characteristics with WWS. An MEB case series [quoted within Ref. 1] describes repeated general anesthesia with volatiles and succinylcholine causing severe increases in CPK levels. We, accordingly, forwent possible Malignant Hyperpyrexia triggers by utilizing propofol based TIVA, despite the uneventful first surgery on volatiles

As we show above – what seemed as a possible anesthetic plan the first time can mean possible harm a second one. Previous case reports of WWS were uninformative as to a 'second hit' effect of anesthetic exposure, post-diagnosis. Our learning point from the case is that the SAC genetic status should be verified *ad hoc* preoperatively, as it entails important and crucial considerations regarding the safety and efficacy of anesthetic management perioperatively.

Acknowledgments

The report is published with the written assent of the patient's parents.

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