RAFFAELE FALSAPERLA
VALENTINA GIACCHI

I rischi del neonato quando arriva a casa: DAL NIDO A CASA

Azienda Policlinico

PO SAN MARCO

Catania

Roadmap

- ✓ Neonato “critico”
- ✓ Stratificazione del rischio neonato in 3 classi
- ✓ Neonato “apparentemente sano” dal centro nascita

Patologie cardiache
Patologie metaboliche
Patologie ad insorgenza neurologica

Neonato critico

Grave compromissione clinica con alterazione dei parametri vitali: **attività respiratoria, circolo metabolismo, temperatura corporea, livello di coscienza**

ELSEVIER

Medicine

Evaluation and Management of the Critically Ill Neonate in the Emergency Department

U. Olivia Kim, MD*, David C. Brousseau, MD MS†, G. Ganesh Konduri, MD*

Table 2 A mnemonic for the differential diagnoses of a critically ill neonate: NEO SECRETS.

N	iNborn errors of metabolism
E	Electrolyte abnormalities
O	Overdose (toxin, poison)
S	Seizures
E	Enteric emergencies
C	Cardiac abnormalities
R	Recipe (formula, herbs, additives)
E	Endocrine crisis
T	Trauma
S	Sepsis

- *Molte cause*
- *Pochi segni*
- *Sintomi aspecifici*

	Signs and Symptoms	Diagnostic Workup
Hyponatremia (Na^+ <130 mEq/L)	Edema Vomiting Muscle weakness Seizures and/or coma	If ↑ K and shock, r/o CAH If ↓UOP and ↓serum osmolality, r/o SIADH or renal defect If ↑ UOP and dehydration, r/o improper mixing of formula
Hypernatremia (Na^+ >150 mEq/L)	Lethargy or irritability Seizures, decreased tone	If ↑ UOP and ↓urine specific gravity, r/o renal concentration defects If ↓UOP and ↑specific gravity, r/o breastfeeding failure, improper formula mixing
Hypokalemia (K^+ <3 mEq/L)	Weakness Ileus Hypertension ECG changes: ↑ P wave amplitude, inverted T waves	If ↑ urine K, r/o renal losses If hypertensive, r/o excess mineralocorticoids or renin If normotensive, r/o RTA, diuretics, GI losses or malnutrition
Hyperkalemia (K^+ > 6 mEq/L)	Weakness Tetany ECG changes: peaked T waves, wide QRS, ST segment depression, V-fib	If ↓urine K+ and ↑ 17-OHP, r/o CAH If ↓UOP, r/o renal failure If ↑ chloride and metabolic acidosis, r/o type IV RTA R/o metabolic acidosis because this can cause factitious hyperkalemia
Hypocalcemia (total serum Ca^{2+} <7 mg/dL, ionized Ca^{2+} <1.1 mmol/L)	Clonic seizures, jerking or tetany Laryngospasm, stridor, weak cry Prolonged QT interval	If ↓Mg+ correct Mg first If ↑ serum Phos, r/o high Phos formula or cow's milk ingestion, ↓PTH If ↑ serum Phos, ↑ urine Ca, r/o renal failure If ↓PTH, ↓Phos, ↑ AP, r/o Vit D abnormality If ↑ PTH, ↓Phos, ↑ AP, r/o rickets If no thymus on CXR, r/o DiGeorge syndrome
Hypoglycemia (serum glucose <40 mg/dL)	Lethargy Seizures	Often symptom of other illness (sepsis, asphyxia, shock, metabolic disorder) If ↑ insulin ↓insulin-like growth factor 1, r/o hyperinsulinemia

r/o indicates rule out; UOP, urine output; SIADH, syndrome of inappropriate antidiuretic hormone; DTRs, deep tendon reflexes; RTA, renal tubular acidosis; ECG, electrocardiogram; V-fib, ventricular fibrillation; GI, gastrointestinal; 17-OHP, 17-hydroxyprogesterone; Ca, calcium; Mg, magnesium; Phos, phosphorus; PTH, parathyroid hormone; AP, alkaline phosphatase; Vit D, vitamin D; K, potassium; CAH, congenital adrenal hyperplasia; Na, sodium; CXR, chest x-ray.

Evaluation and Management of the Critically Ill Neonate in the Emergency DepartmentClin Ped Emerg Med 9:140-148

ORIGINAL ARTICLE
Emergency & Critical Care Medicine

JKMS

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The Clinical Differences between Urgent Visits and Non-Urgent Visits in Emergency Department During the Neonatal Period

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As neonates are brought to the emergency department (ED) for various complaints, it is challenging for emergency physicians to clinically determine the urgency of the visit. We sought to explore clinical characteristics associated with urgent visits to the ED. We conducted a retrospective study by reviewing medical records of neonatal visits to a tertiary pediatric regional emergency center for 5 years. Cases of patients who were discharged after checking only chest or abdominal X-ray or discharged without workup, were classified as non-urgent visits. Cases where more examinations were performed, or when the patient was hospitalized, were classified as urgent visits. Various clinical features and process in the ED were compared between the groups. Of the 1,008 cases enrolled in this study, 856 (84.9%) were urgent and 152 (15.1%) were non-urgent visits. After adjustment by multiple logistic regression analysis, non-urgent visits were associated with self-referrals rather than physician-referrals (odds ratio [OR], 5.96), visits in the evening rather than at night or daytime (OR, 2.51), patient visits from home rather than from medical facilities (OR, 2.19; 95). Fever and jaundice were the most common complaints (25.7% and 24.5%, respectively), and their OR of non-urgent visit was relatively low (adjusted OR 0.03 and 0.03 ,respectively). However, other common complaints, such as vomiting and cough (7.4% and 7.1%, respectively), were more likely to be non-urgent visits (adjusted OR 2.96 and 9.83, respectively). For suspected non-urgent visits, emergency physicians need to try to reduce unnecessary workup and shorten length of stay in ED.

Keywords: Neonate; Emergency Department; Urgent; Non-urgent

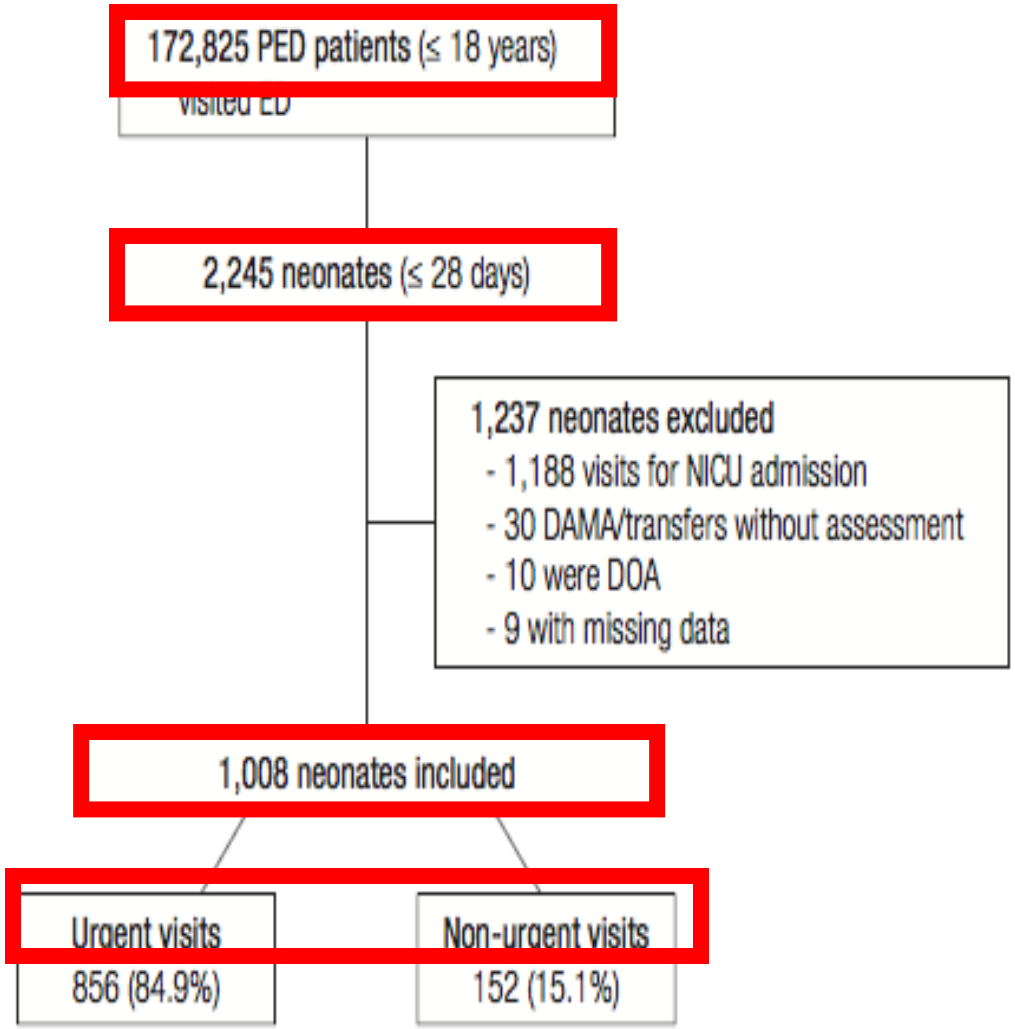


Fig. 1. Flow chart of patient inclusion.
PED = pediatric emergency department, NICU = neonatal intensive care unit, DAMA = discharge against medical advice, DOA = dead on arrival.

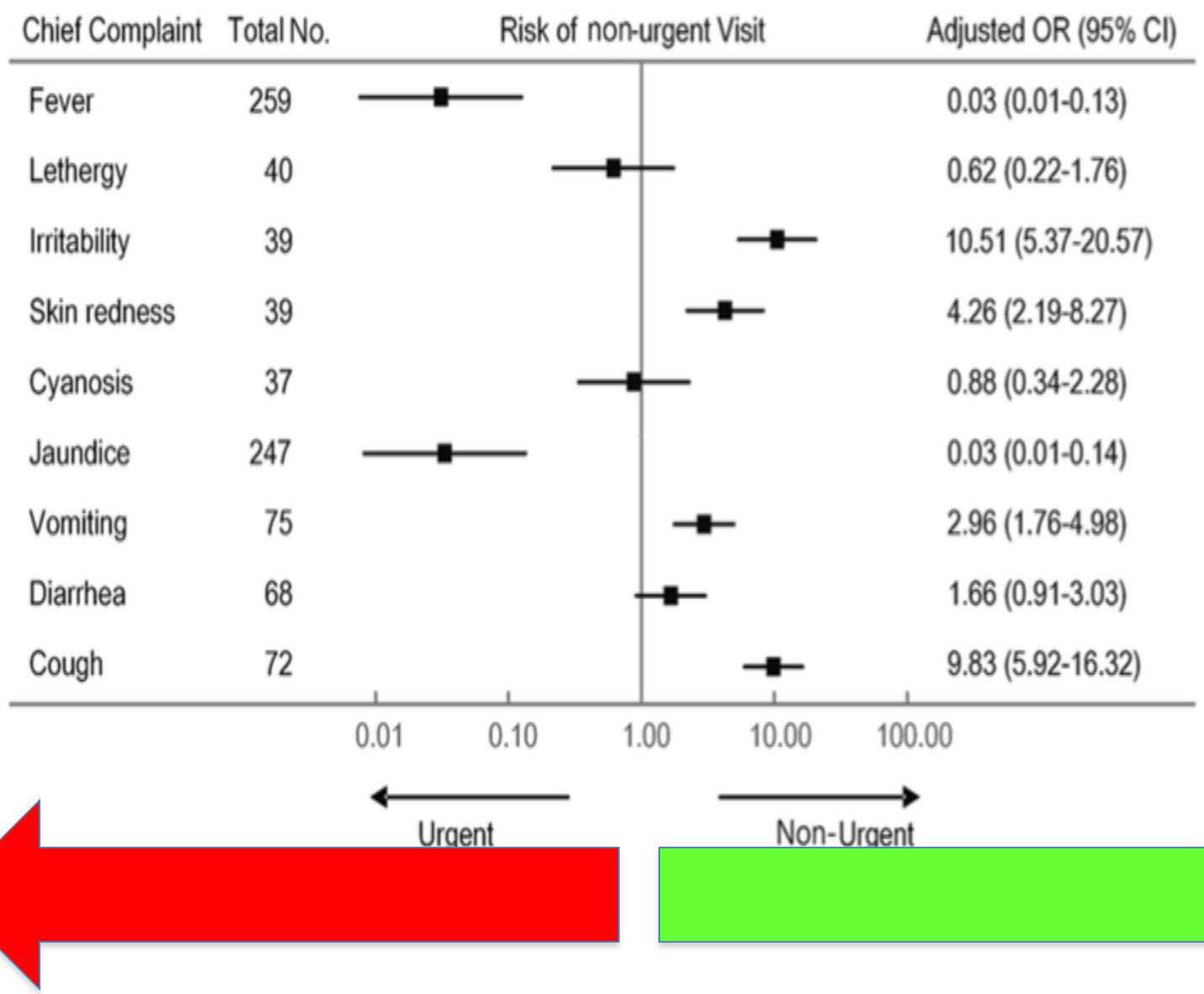
Table 3. Presentations by urgent/non-urgent visits (from most to least common)

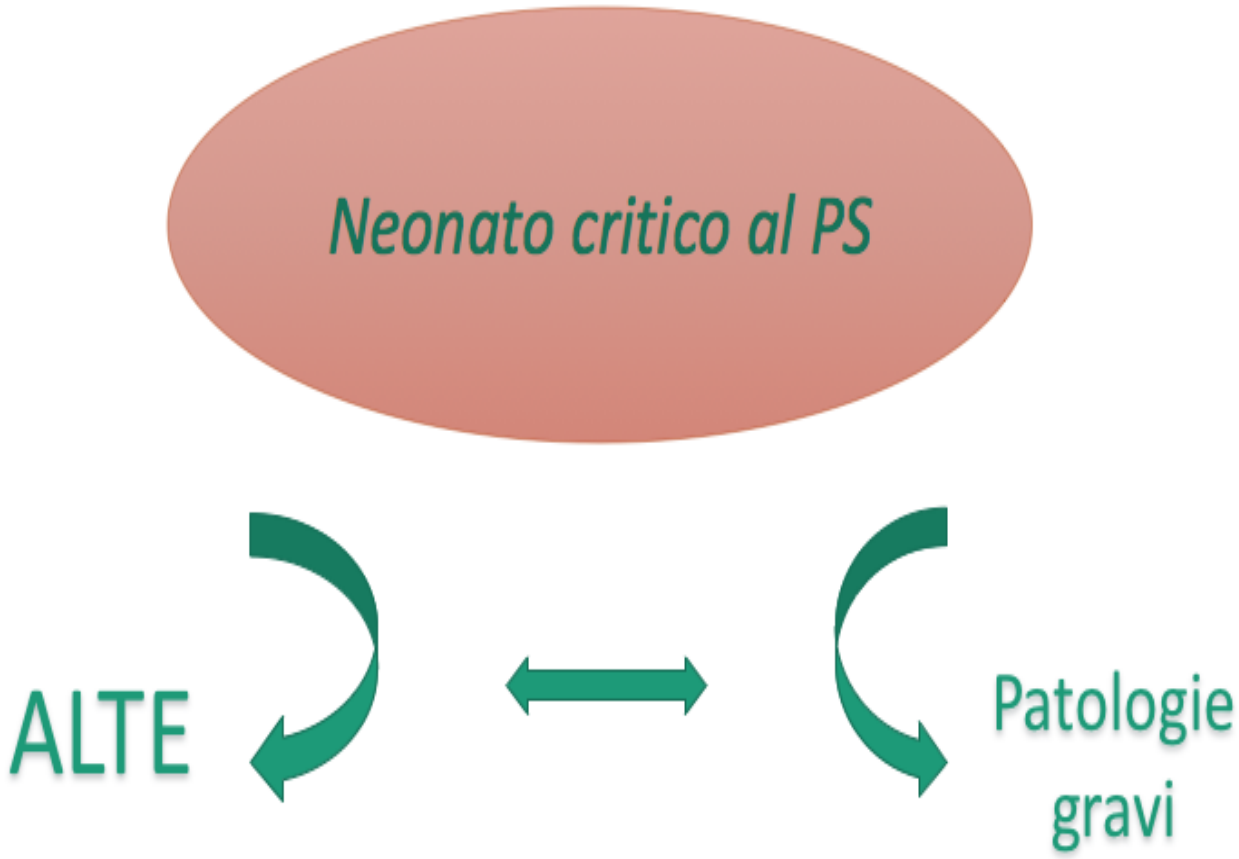
Chief complaints	Total (n = 1,008)	Urgent (n = 856)	Non-urgent (n = 152)
General appearance abnormalities	416 (41.3)	364 (42.5)	52 (34.2)
Fever	259 (25.7)	257 (30.0)	2 (1.3)
Lethargy	40 (4.0)	36 (4.2)	4 (2.6)
Irritability	39 (3.9)	15 (1.8)	24 (15.8)
Skin redness	39 (3.9)	23 (2.7)	16 (10.5)
Cyanosis	37 (3.7)	32 (3.7)	5 (3.3)
Premature	2 (0.2)	1 (0.1)	1 (0.7)
Gastrointestinal complaints	421 (41.8)	374 (43.7)	47 (30.9)
Jaundice	247 (24.5)	245 (28.6)	2 (1.3)
Vomiting	75 (7.4)	51 (6.0)	24 (15.8)
Diarrhea	68 (6.7)	53 (6.2)	15 (9.9)
Hematochezia	19 (1.9)	18 (2.1)	1 (0.7)
Abdominal distension	8 (0.8)	4 (0.5)	4 (2.6)
Hematemesis	4 (0.4)	3 (0.4)	1 (0.7)
Respiratory complaints	97 (9.6)	51 (6.0)	46 (30.3)
Cough	72 (7.1)	31 (3.6)	41 (27.0)
Tachypnea	13 (1.3)	11 (1.3)	2 (1.3)
Respiratory distress	12 (1.2)	9 (1.1)	3 (2.0)
Cardiologic complaints	9 (0.9)	8 (0.9)	1 (0.7)
Cardiac murmur	8 (0.8)	7 (0.8)	1 (0.7)
Bradycardia	1 (0.1)	1 (0.1)	0 (0.0)
Neurologic complaints	25 (2.5)	25 (2.9)	0 (0.0)
Seizure	25 (2.5)	25 (2.9)	0 (0.0)
Congenital complaints	9 (0.9)	9 (1.1)	0 (0.0)
Congenital abnormality	9 (0.9)	9 (1.1)	0 (0.0)
Others	31 (3.1)	25 (2.9)	6 (3.9)
Hematoma, fracture	13 (1.3)	10 (1.2)	3 (2.0)
Scrotal swelling	7 (0.7)	6 (0.7)	1 (0.7)
Hematuria	6 (0.6)	4 (0.5)	2 (1.3)
Abnormal laboratory results	4 (0.4)	4 (0.5)	0 (0.0)
Intoxication	1 (0.1)	1 (0.1)	0 (0.0)

Values are presented as number (%).

Diagnosis	Urgent (n = 856)	Non-urgent (n = 152)
Infection	444 (51.9)	84 (55.3)
Acute gastroenteritis	153 (17.9)	25 (16.4)
Sepsis	152 (17.8)	0 (0.0)
Acute pyelonephritis	32 (3.7)	11 (7.2)
Pneumonia	22 (2.6)	4 (2.6)
Bronchiolitis	22 (2.6)	6 (3.9)
Skin infection	21 (2.5)	12 (7.9)
Meningitis	19 (2.2)	1 (0.7)
Upper respiratory infection	12 (1.4)	23 (15.1)
Other infection	11 (1.3)	2 (1.3)
Gastrointestinal problem	287 (33.5)	34 (22.4)
Hyperbilirubinemia	251 (29.3)	3 (2.0)
Intestinal bleeding	13 (1.5)	3 (2.0)
Dehydration	10 (1.2)	1 (0.7)
Ileus	9 (1.1)	5 (3.3)
Infantile colic	2 (0.2)	15 (9.9)
Constipation	1 (0.1)	7 (4.6)
Gastroesophageal reflux disease	1 (0.1)	0 (0.0)
Congenital anomaly	32 (3.7)	1 (0.7)
Cardiologic problem	22 (2.6)	2 (1.3)
Congenital heart disease	20 (2.4)	2 (1.3)
Arrhythmia	2 (0.2)	0 (0.0)
Normal newborn	8 (0.9)	18 (11.8)
Respiratory problem	15 (1.8)	7 (4.6)
Aspiration	6 (0.7)	5 (3.3)
Transient tachypnea	5 (0.6)	1 (0.7)
Pneumothorax	2 (0.2)	0 (0.0)
Apnea	2 (0.2)	1 (0.7)
Neurologic problem	19 (2.2)	0 (0.0)
Seizure	18 (2.1)	0 (0.0)
Cerebral infarction	1 (0.1)	0 (0.0)
Endocrinologic/nephrologic/hematologic problem	16 (1.9)	2 (1.3)
Hypocalcemia	8 (0.9)	0 (0.0)
Hypoglycemia	2 (0.2)	0 (0.0)
Dysuria	2 (0.2)	1 (0.7)
Hypothyroidism	2 (0.2)	0 (0.0)
Anemia	1 (0.1)	0 (0.0)
Hydrocele	1 (0.1)	1 (0.7)
Trauma/toxic problem	13 (1.5)	4 (2.6)
Contusion and fracture	10 (1.2)	4 (2.6)
Poisoning	3 (0.3)	0 (0.0)

Values are presented as number (%).


Chief Complaint
Total No.
Risk of non-urgent Visit
Adjusted OR (95% CI)
Fever
259
0.03 (0.01-0.13)
Lethergy
40
0.62 (0.22-1.76)
Irritability
39
10.51 (5.37-20.57)
Skin redness
39
4.26 (2.19-8.27)
Cyanosis
37
0.88 (0.34-2.28)
Jaundice
247
0.03 (0.01-0.14)
Vomiting
75
2.96 (1.76-4.98)
Diarrhea
68
1.66 (0.91-3.03)
Cough
72
9.83 (5.92-16.32)
0.01
0.10
1.00
10.00
100.00
Urgent
Non-Urgent



Quali esami si rendono utili in PS per discriminare un ALTE isolato da eventi ricorrenti che sottendono patologie più gravi?

Causes of apparent life threatening events in infants: a systematic review

M C McGovern, M B H Smith

Arch Dis Child 2004;**89**:1043–1048. doi: 10.1136/adc.2003.031740

- ***ALTE (Apparent Life-Threatening Event):*** *episodio spaventoso per l'osservatore caratterizzato da una associazione di* ***apnea*** *(centrale o occasionalmente ostruttiva),* ***alterazioni del colorito*** *(cianotico, pallido, eritematoso, pletorico), alterazioni del tono muscolare* ***(ipotonia),*** *soffocamento o conati di vomito (National Institute of Health Consensus Conference on Infantile Apnea 1986)*
- ***Tali eventi solo raramente sono una manifestazione di un quadro patologico grave***

THE JOURNAL OF PEDIATRICS • www.jpeds.com

Management of Apparent Life-Threatening Events in Infants: A Systematic Review

Joel S. Tieder, MD, MPH[1], Robin L. Altman, MD[2], Joshua L. Bonkowsky, MD, PhD[3], Donald A. Brand, PhD[4,5], Ilene Claudius, MD[6], Diana J. Cunningham, MLS, MPH, AHIP[7], Craig DeWolfe, MD, Med[8], Jack M. Percelay, MD, MPH[9], Raymond D. Pitetti, MD, MPH[10], and Michael B. H. Smith, MB, FRCPC, FRCPCH[11]

Objective To determine in patients who are well-appearing and without a clear etiology after an apparent life-threatening event (ALTE): (1) What historical and physical examination features suggest that a child is at risk for a future adverse event and/or serious underlying diagnosis and would, therefore, benefit from testing or hospitalization? and (2) What testing is indicated on presentation and during hospitalization?

Study design Systematic review of clinical studies, excluding case reports, published from 1970 through 2011 identified using key words for ALTE.

Results The final analysis was based on 37 studies; 18 prospective observational, 19 retrospective observational. None of the studies provided sufficient evidence to fully address the clinical questions. Risk factors identified from historical and physical examination features included a history of prematurity, multiple ALTEs, and suspected child maltreatment. Routine screening tests for gastroesophageal reflux, meningitis, bacteremia, and seizures are low yield in infants without historical risk factors or suggestive physical examination findings.

Conclusion Some historical and physical examination features can be used to identify risk in infants who are well-appearing and without a clear etiology at presentation, and testing tailored to these risks may be of value. The true risk of a subsequent event or underlying disorder cannot be ascertained. A more precise definition of an ALTE is needed and further research is warranted. *(J Pediatr 2013;■:■-■).*

Neonati a rischio

- Anomalie congenite
- Trauma da parto
- Prematurità
- Neonati piccoli per l'età gestazionale
- Apgar score < 7
- Ospedalizzazione

Türk Pediatri Arşivi
Turkish Archives of Pediatrics

Turkish Neonatal Society guideline for the follow-up of high-risk newborn infants

Türk Neonatoloji Derneği yüksek riskli bebek izlem rehberi

Betul Acunaş[1], Sinan Uslu[2], Ahmet Yağmur Baş[3]

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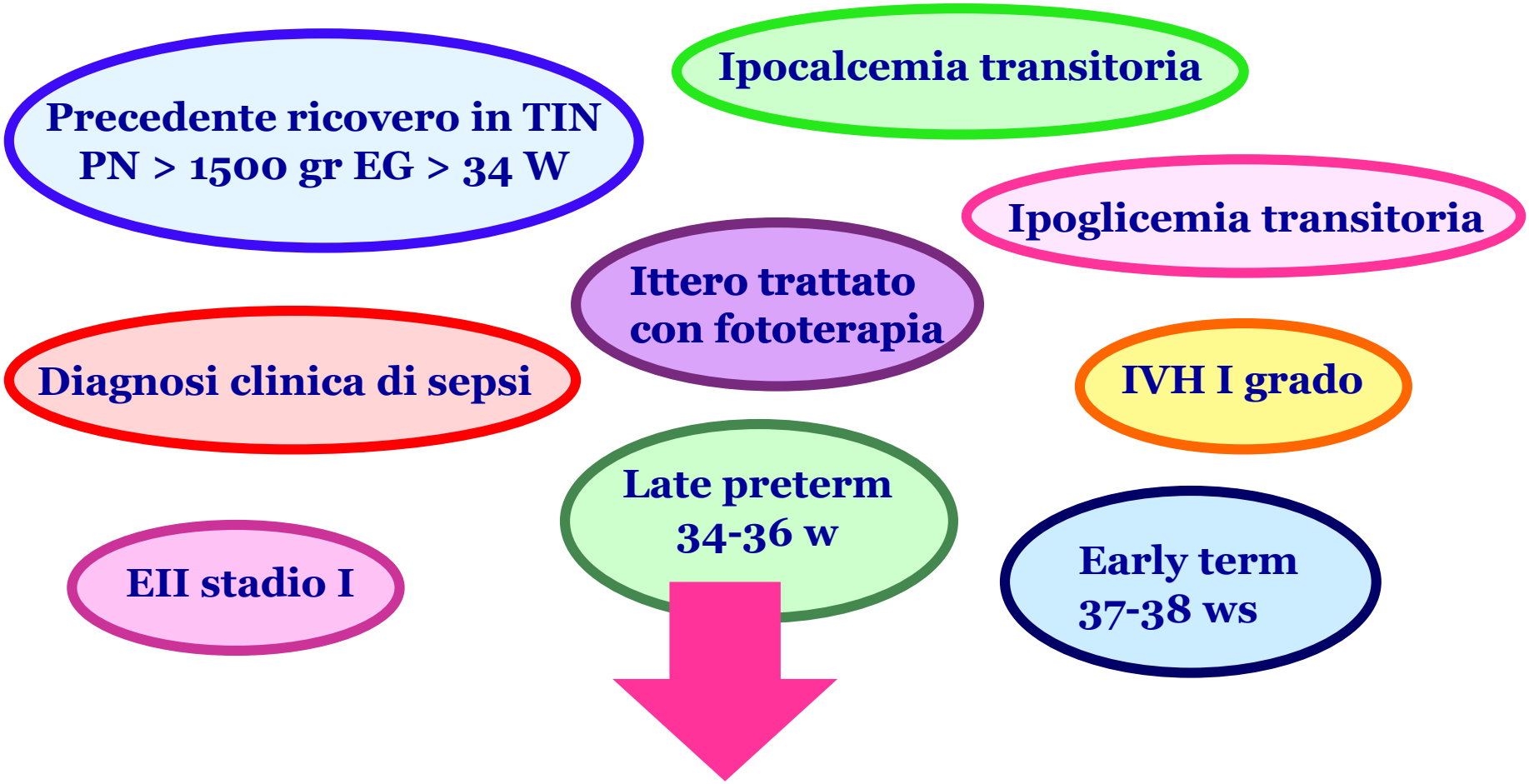
[3]Division of Neonatology, Department of Pediatrics, Yıldırım Beyazıt University, Faculty of Medicine, Ankara, Turkey

Table 1. Risk categories

	High-risk (Level 3–4)	Moderate risk (Level 2)	Mild risk (Level 1)
Gestation week and birth weight	<1000 g <29 wk SGA and LGA babies Babies with fetal malnutrition	1000-1500 g 29–34 weeks Multiple pregnancies	Hospitalized in NICU with a birth weight of >1500 g and a gestational age of >34 wks Late preterm Early term
Central nervous system	Stage 3-4 ICH Ventriculomegaly Cystic PVL Hydrocephaly Perinatal asphyxia[a] Convulsion Cerebral infarction Abnormal neurologic examination findings at the time of discharge Need for advancedresuscitation at birth	Stage 2 ICH Stage 2 HIE Need for basic resuscitation at birth	Stage 1 ICH Stage 1 HIE
Respiratory system	Prolonged (>7 days) MV High frequency ventilation BPD Pneumothorax Severe apnea	MV support (<7 days) CPAP application	
Cardiovascular system	ECMO-iNO application PPH Shock requiring inotropic agents Severe thrombosis	Catheter placement (umbilical central catheter or peripherally inserted central catheter)	
Infectious conditions	Severe sepsis Meningitis Nosocomial infection Baby of HIV-positive mother Intrauterine infections	Sepsis confirmed with culture	Clinical sepsis
Surgical problems	Diaphragm hernia Tracheo-esophageal fistula Duodenal atresia Surgical NEC PDA with surgical ligation Laser applied ROP Cardiac surgical diseases Shunt operated hydrocephaly	NEC and PDA requiring medical treatment	Other surgical interventions (e.g. inguinal, umbilical hernia)
Other	Prolonged hypoglycemia Prolonged hypocalcemia Twin-to-twin transfusion Jaundice requiring exchange transfusion Bilirubin encephalopathy Major congenital malformation Metabolic/genetic diseases	Hypoglycemia (≤25 mg/dL, >3 days) Severe jaundice Partial exchange transfusion Inappropriate environmental conditions[b]	Transient hypoglycemia Hypocalcemia Jaundice requiring phototherapy

BPD: bronchopulmonary dysplasia;
CPAP: continuous positive airway pressure; ECMO: extracorporeal membrane oxygenation;
HIE: hypoxic ischemic encephalopathy;
ICH: intracranial hemorrhage;
iNO: inhaled nitric oxide;
LGA: large-for-gestational-age baby;
MV: mechanical ventilation;
NEC: necrotizing enterocolitis;
SGA: small-for-gestational-age baby;
PDA: patent ductus arteriosus;
ROP: retinopathy of prematurity;
PPH: persistent pulmonary hypertension; PVL: periventricular leukomalacia

Neonati a basso rischio



FOLLOW-UP DAL PEDIATRA DI BASE

Acunaş B, Uslu S, Yağmur Baş A. *Turkish Neonatal Society guideline for the follow-up of high-risk newborn infants*. Turk Pediatri Ars 2018; 53(Suppl 1): S180-S195.

DIMISSIONE PRECOCE < 48 ORE DEI NEONATI A TERMINE APPARENTEMENTE SANI

L'American Academy of Paediatrics raccomanda un controllo clinico entro 2-3 giorni dalla dimissione

Lutfi S, Al-Rifai H, Al-Ansari K. Neonatal visits to the pediatric emergency center and its implications onpostnatal discharge practices in Qatar. J Clin Neonatol 2013;2:14-9.

PLOS ONE

RESEARCH ARTICLE

The prevalence of adverse postnatal outcomes for mother and infant in the Netherlands

Nynke de Groot[1]*, Erwin Birnie[1], Jolanda H. Vermolen[2], Jacqueline J. A. Dorscheidt[2], Gouke J. Bonsel[1]

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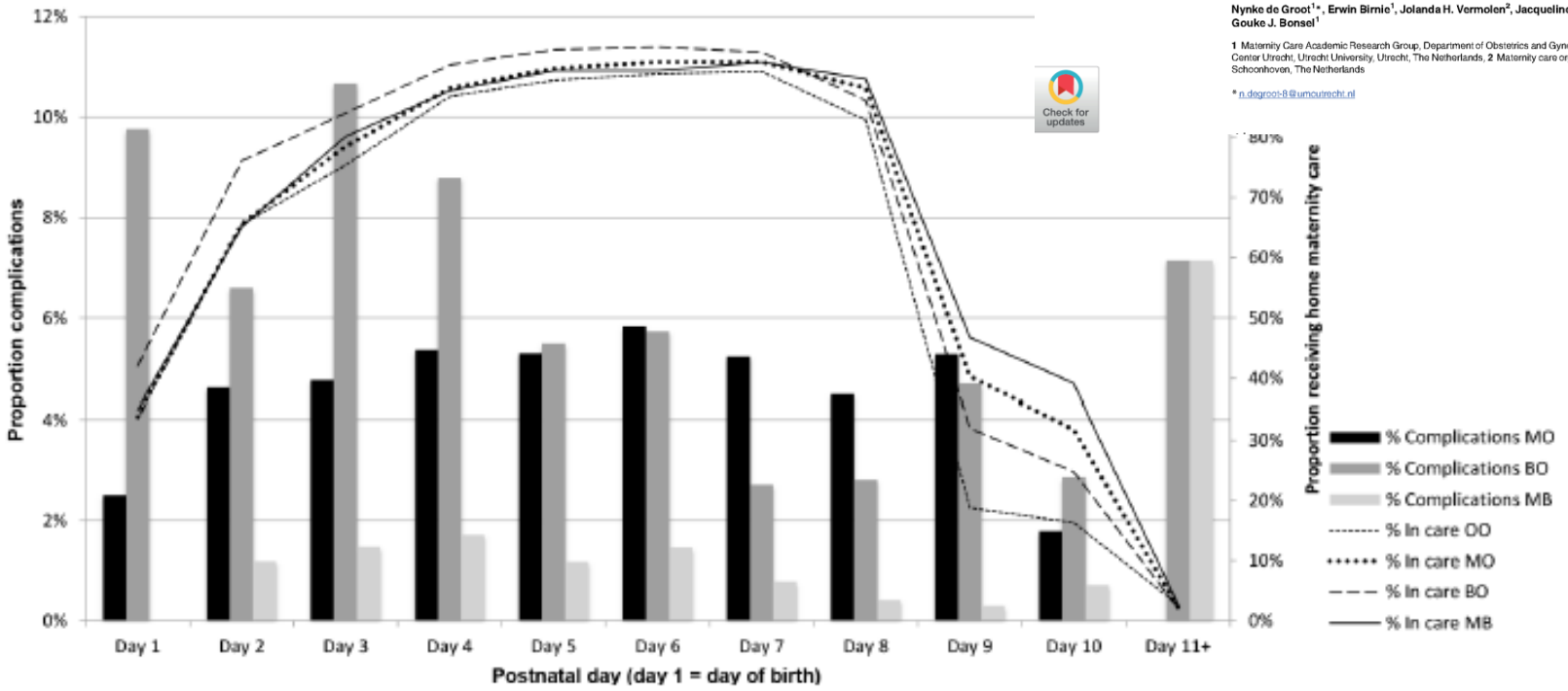

Fig 1. Daily postnatal complications rate (bars) and proportion of mother-baby pairs receiving home maternity care (lines) by postnatal day.

https://doi.org/10.1371/journal.pone.0202960.g001

de Groot N, Birnie E, Vermolen JH, Dorscheidt JJA, Bonsel GJ (2018) *The prevalence of adverse postnatal outcomes for mother and infant in the Netherlands*. PLoS ONE 13(9):e0202960. *https://doi.org/10.1371/journal.pone.0202960*

PLOS ONE

RESEARCH ARTICLE

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PLOS ONE

RESEARCH ARTICLE

Maternal detection of neonatal jaundice during birth hospitalization using a novel two-color icterometer

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Check for updates

Abstract

Hyperbilirubinemia, manifesting as jaundice, affects about 60–80% of all newborns and is a leading cause of hospital admission or re-hospitalization in the first week of life [1–3]. Mothers are frequently the first to observe jaundice in the vast majority of the affected newborns, especially outside hospital settings [4,5]. Maternal role is even of more interest in low-and middle-income countries with a high prevalence of risk factors for neurotoxicity and a high proportion of deliveries outside hospitals [6]. However, poor recognition of jaundice and delayed presentation to hospital remain major contributors to the burden of bilirubin encephalopathy, excessive rates of avoidable exchange transfusion and related adverse outcomes [1,7]. Maternal education and empowerment for timely detection of jaundice is therefore, crucial for effective community-based interventions needed to curtail the burden of severe neonatal jaundice [8–10].

Table 1: Admission diagnosis for neonates re-hospitalized after postnatal discharge

Reason for admission	Number 61 (100%)
Bronchiolitis	19 (31)
Neonatal sepsis	14 (23)
Apnea/seizures	7 (11)
Jaundice	5 (8)
Suspected congenital heart disease	3 (5)
Dermatological condition/skin infection	3 (5)
Urinary tract infection	3 (5)
Vomiting	2 (3)
Others	5 (8)

Lutfi S, Al-Rifai H, Al-Ansari K. Neonatal visits to the pediatric emergency center and its implications onpostnatal discharge practices in Qatar. J Clin Neonatol 2013;2:14-9.

EMERGENZE AD INSORGENZA NEONATALE

PATOLOGIE INFETTIVE

Sepsi

PATOLOGIE NEUROMUSCOLARI

Distrofie muscolari congenite

PATOLOGIE NEUROLOGICHE

Convulsioni

PATOLOGIE METABOLICHE

Citrullinemia
Argininsuccinico aciduria
Deficit di Crotonil CoA carbossilasi
Deficit di biotinidasi
Galattosemia variante Duarte
Acidemia propionica
Acidemia isovalerica
Acidemia malonica
MCAD
SCAD

PATOLOGIE ENDOCRINOLOGICHE

Ipotiroidismo
Ipoparatiroidismo

LONS
Sepsi neonatale a insorgenza tardiva
(*late-onset neonatal sepsis*)

- **esordio dopo le prime 48-72 ore di vita;**
- **0,3-3% dei neonati a termine;**
- **6,2-33% di tutti i neonati ricoverati in UTIN;**
- **Età media al primo episodio: 2-3 settimane**

SIN
SOCIETÀ ITALIANA DI NEONATOLOGIA
Gruppo di Studio di Infettivologia Neonatale

LONS
Sepsi neonatale a insorgenza tardiva
(*late-onset neonatal sepsis*)

- **Quasi sempre microorganismi provenienti dall'ambiente**

Compito del pediatra: sottolineare l'importanza del rispetto delle norme igienico sanitarie e del lavaggio delle mani

- **Talora dal tratto urogenitale materno**

SIN
SOCIETÀ ITALIANA DI NEONATOLOGIA
Gruppo di Studio di Infettivologia Neonatale

LONS: CAMPANELLI D'ALLARME

Suzione poco valida o assente
pianto flebile o assente
ipotonia muscolare agli arti o generalizzata
vomito o distensione addominale
cute fredda, marezzata
rientramenti del torace
Precedente ricovero in UTIN
TVR materno positivo per GBS

RICOVERO IN OSPEDALE

LONS

- Streptococco β emolitico di gruppo B: spesso forme a decorso fulminante
- batteri Gram-negativi: spesso shock settico da endotossinemia (lipopolisaccaride, LPS);
- CoNS: decorso più spesso subdolo.

Disordini parossistici non epilettici

TREMORI

- Coinvolgimento simmetrico delle estremità che risparmia i muscoli facciali
- Movimenti di uguale frequenza ed ampiezza
- Frequenza maggiore rispetto ai movimenti clonici
- Spesso scatenati da uno stimolo
- EII, ipocalcemia, ipoglicemia, astinenza da farmaci
- Anche in bambini senza coinvolgimento neurologico

David Evans, Malcolm Levene*Arch Dis Child Fetal Neonatal Ed* 1998;**78**:F70–F75

COARTAZIONE AORTICA

- **Incidenza: 3/10000 nati vivi**

Cangussù et al. Rev Assoc Med Bras (1992). 2019

- **Incidenza fetale 6-7 % di tutte le cardiopatie congenite**

- **Incidenza neonatale 5-6 % di tutte le cardiopatie congenite**

Lindsey D Allan et al. JACC 1994

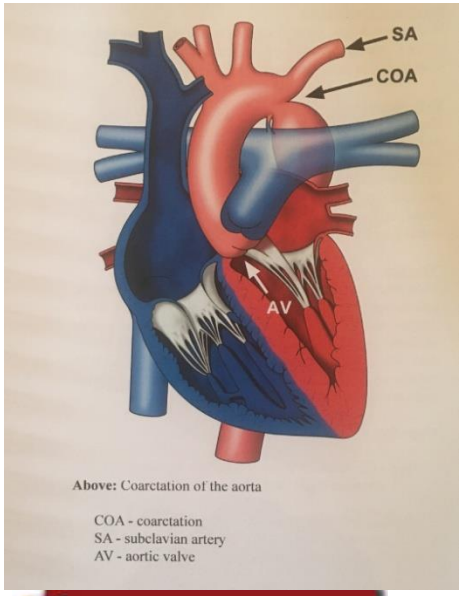
Ferencz et al. 1993

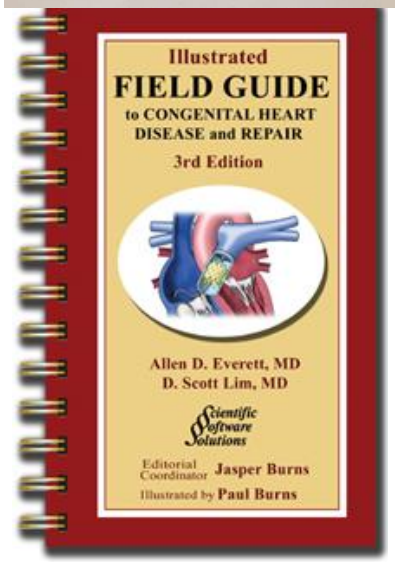
- **Il 35 % delle bambine affette da S. di Turner presentano coartazione dell'aorta**

Trimble AK et al. 1998

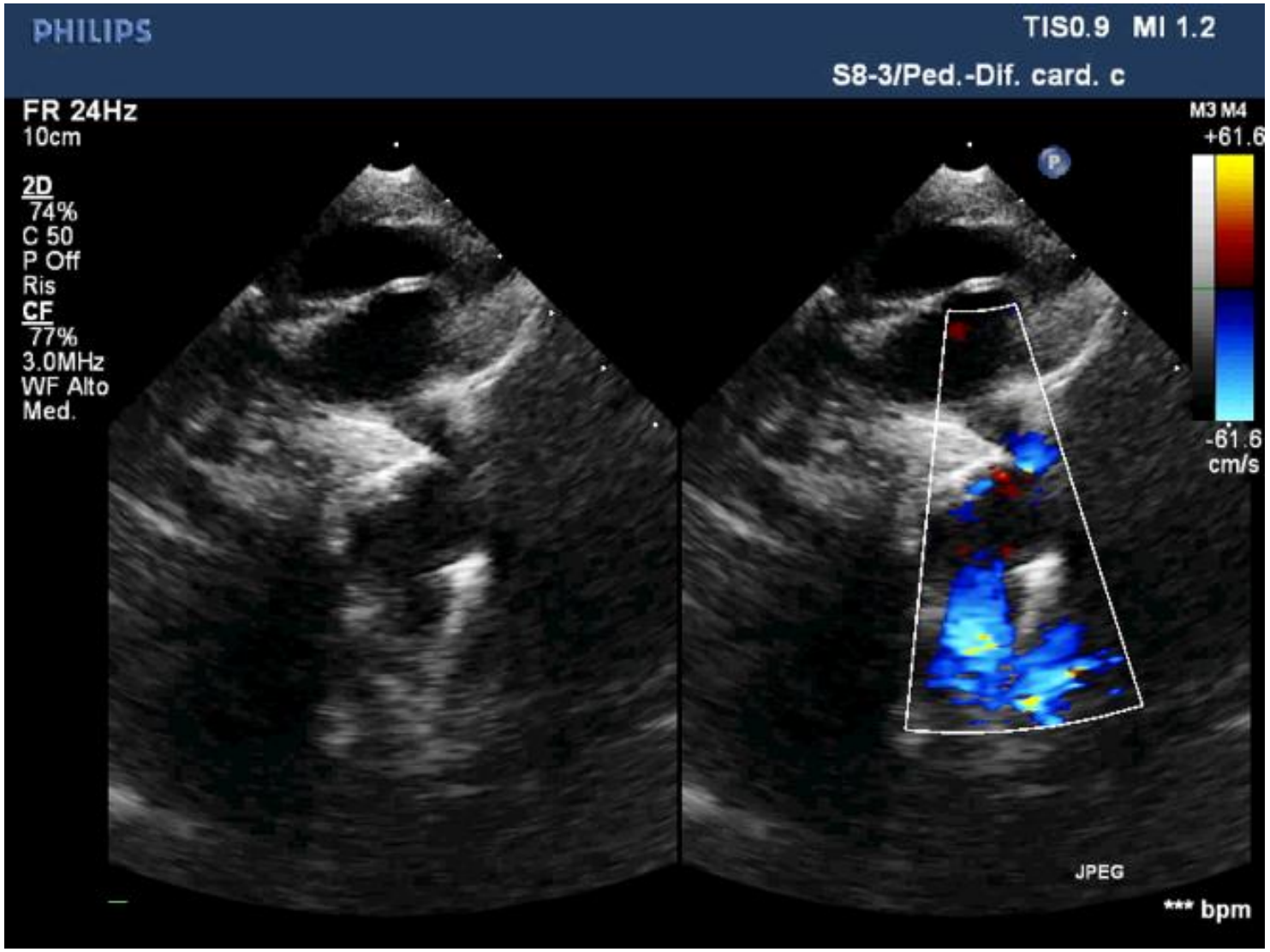
COARTAZIONE AORTICA

- Restringimento del lume dell'aorta dovuta ad uno shelf posteriore ostruttivo in regione iuxtaduttale o ad ipoplasia tubulare completa.
- Nel caso di shelf posteriore il mantenimento del Dotto di Botallo pervio può limitare il grado di ostruzione
- Meno comunemente coinvolti l'arco aortico trasverso o l'aorta addominale
- Possibile associazione con DIV, anello mitrale sopravalvolare, stenosi sottoaortica



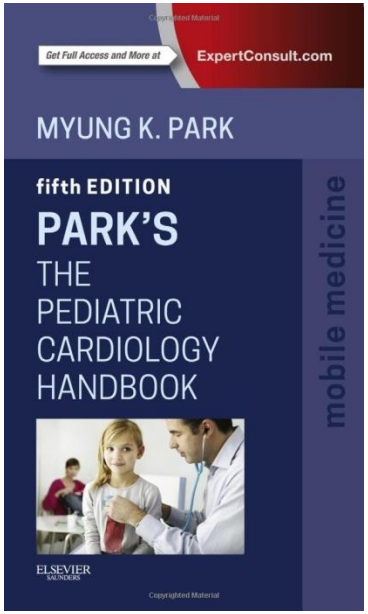


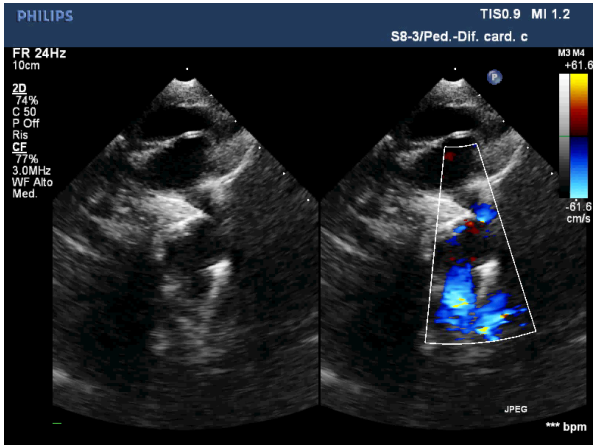
COARTAZIONE AORTICA



COARTAZIONE AORTICA
VALUTAZIONE

- Cianosi
- Tempo di Refill > 2’’
- Polsi femorali
- SpO2 pre e post duttale
- Tachicardia (FC > 180 bpm)
- Dispnea
- Difficoltà di suzione
- Oligo-anuria
- Ritmo di galoppo
- PA arti superiori – arti inferiori





Utilità del saturimetro nello screening delle cardiopatie non cianogene

Può un neonato portatore di una cardiopatia congenita essere dimesso dal nido senza diagnosi?

Cardiopatie congenite che più frequentemente sfuggono alla diagnosi: CoA (60%) TAVPR (15%)

Utilità del saturimetro nello screening della Coartazione aortica

Un test positivo per screening CHD deve soddisfare uno dei 3 seguenti criteri:

- SaO2 <90 %
- SaO2 <95 % in sia all'arto superiore che inferiore, in 2 misurazioni, a distanza di 1 ora.
- SaO2 differenza >2SaO2 2--3 % tra mano/polso destro e piede sinistro

SCREENING METABOLICO ALLARGATO

Screening Neonatale Metabolico Allargato
L. 167/2016 - DA n.579/2017
Centro di riferimento regionale per la prevenzione, la diagnosi e la cura delle malattie rare del metabolismo dell'infanzia
Clinica Pediatrica, AOU Policlinico-Vittorio Emanuele
Catania

Laboratorio di Screening Neonatale Metabolico Allargato
AOU-Policlinico-VE
Ed. 9, piano 1, Lab: 9/01/68-69-70
Via S. Sofia, 78 95123, Catania
095 3781493 – 2886
095 3781493
screeningneonatale@policlinico.unict.it

A cosa servono gli screening neonatali?

Riconoscere come malato un neonato apparentemente sano, tra tanti sani

Screening neonatale metabolico allargato

- Intervento di medicina preventiva Individuazione precoce dei neonati a rischio di malattie congenite:
- Inizio tempestivo iter diagnostico
- Avvio terapia
- Riduzione mortalità, morbilità e grave disabilità Consulenza genetica

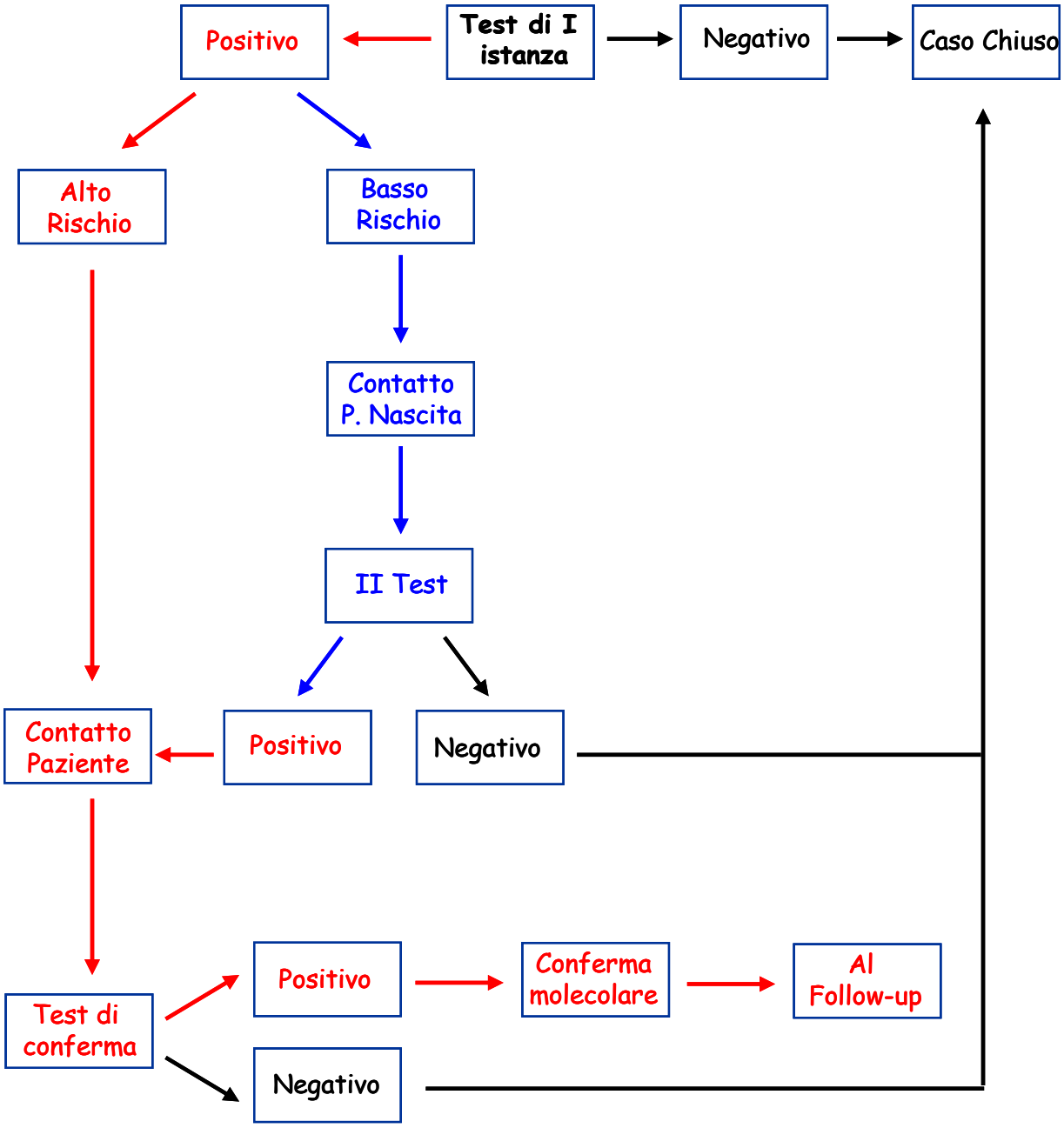
Malattie metaboliche:

- Malattie genetiche ereditarie rare
- Clinica variabile (da scompenso metabolico acuto ad andamento lentamente progressivo)
- Incidenza complessiva: 1/3.000 nati

Screening neonatale metabolico allargato

“Un test per molte malattie” :

- Sulla stessa goccia di sangue è possibile analizzare un’ampia gamma di metaboliti
- Tecnica innovativa: Spettrometria di Massa Tandem
- Individuazione di **oltre 40 errori congeniti del metabolismo**


Positivo
Test di I istanza
Negativo
Caso Chiuso
Alto Rischio
Basso Rischio
Contatto P. Nascita
II Test
Contatto Paziente
Positivo
Negativo
Test di conferma
Positivo
Conferma molecolare
Al Follow-up
Negativo

MALATTIE METABOLICHE AD ESORDIO NELLA PRIMA SETTIMANA DI VITA DIAGNOSTICABILI PRECOCEMENTE CON TANDEM MASS

Citrullinemia
Argininsuccinico aciduria
Deficit di Crotonil CoA carbossilasi
Deficit di biotinidasi
Galattosemia variante Duarte
Acidemia propionica
Acidemia isovalerica
Acidemia malonica
MCAD, SCAD

MALATTIE METABOLICHE AD ESORDIO NELLA PRIMA SETTIMANA DI VITA DIAGNOSTICABILI PRECOCEMENTE CON TANDEM MASS

Suzione ipovalida
Vomito
Scarsa crescita
Sonnolenza
Letargia
Tachipnea
Apnea
Convulsioni
Compromissione epatica
Acidosi metabolica
Ipotonia
Ipostenia
FORME ASINTOMATICHE

CASI CLINICI

VOMITO E LETARGIA

L.G., maschio
Nato alla 37 W + 5 gg di EG da TC per pregresso TC
PN 2980 gr
APGAR 1'/9, 5'/10
Genitori non consanguinei
Buon adattamento alla vita extrauterina
Dimesso in terza giornata di vita dopo avere eseguito Tandem Mass

In quinta giornata di vita
VOMITO e LETARGIA

Emocromo, PCR, funzionalità epatica e renale nella norma
EGA: pH 6.7, HCO3 9.9 mmol/L, BE – 17.5 mmol/L
IPERSODIEMIA

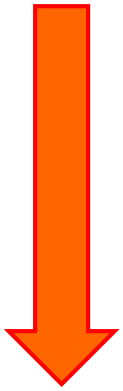
VOMITO E LETARGIA

Ammonio: 761 uM

RISULTATO TANDEM MASS ???

VOMITO E LETARGIA

Elevati livelli di propionilcarnitina (20 Um, V.N. < 6)



ACIDI ORGANICI URINARI: aumento acido 3-idrossi-propionico (160, v.n. < 1.1) e meticitrico (27 v.n. <1.1)

VOMITO E LETARGIA

DIAGNOSI DI ACIDEMIA PROPIONICA

Restrizione proteica, nutrizione ipercalorica, supplementazione con biotina e carnitina

Miglioramento clinico
In corso analisi genetica

UNA "TACHIPNEA SILENTE"

A.S.P., maschio
Nato alla 40 W di EG da parto eutocico
PN 3200 gr
APGAR 1'/9, 5'/10
Genitori non consanguinei
Gravidanza fisiologica
Buono l'adattamento alla vita extrauterina
Dimesso dopo 48 ore: suzione valida al seno materno, alvo regolare, SpO2 pre e post duttale nella norma
Tandem Mass: negativa

A nove giorni di vita
Tachipnea
Difficoltà ad alimentarsi
Restrizione della diuresi

Sospetto contatto con adulto affetto da infezione delle alte vie aeree

UNA "TACHIPNEA SILENTE"

No rinorrea o congestione nasale
Distress respiratorio
No wheezing, ronchi o rantoli
No vomito e/o diarrea
T 37.1 ° C
FC 167 bpm
FR 85 atti/min
PA (braccio destro): 80/55 mmHg
SpO2 95 %
Lieve disidratazione

↓

Emocromo, PCR, elettroliti, esame urine nella norma
Eseguita emocoltura ed urinocoltura
Tampone nasale per virus negativo
EGA: pH 7.3; pCO2 38 mmHg; pO2 102 mmHg;I HCO3 17 mmol/L; BE – 9.9 mmol/L
Rx torace: ndr

↓

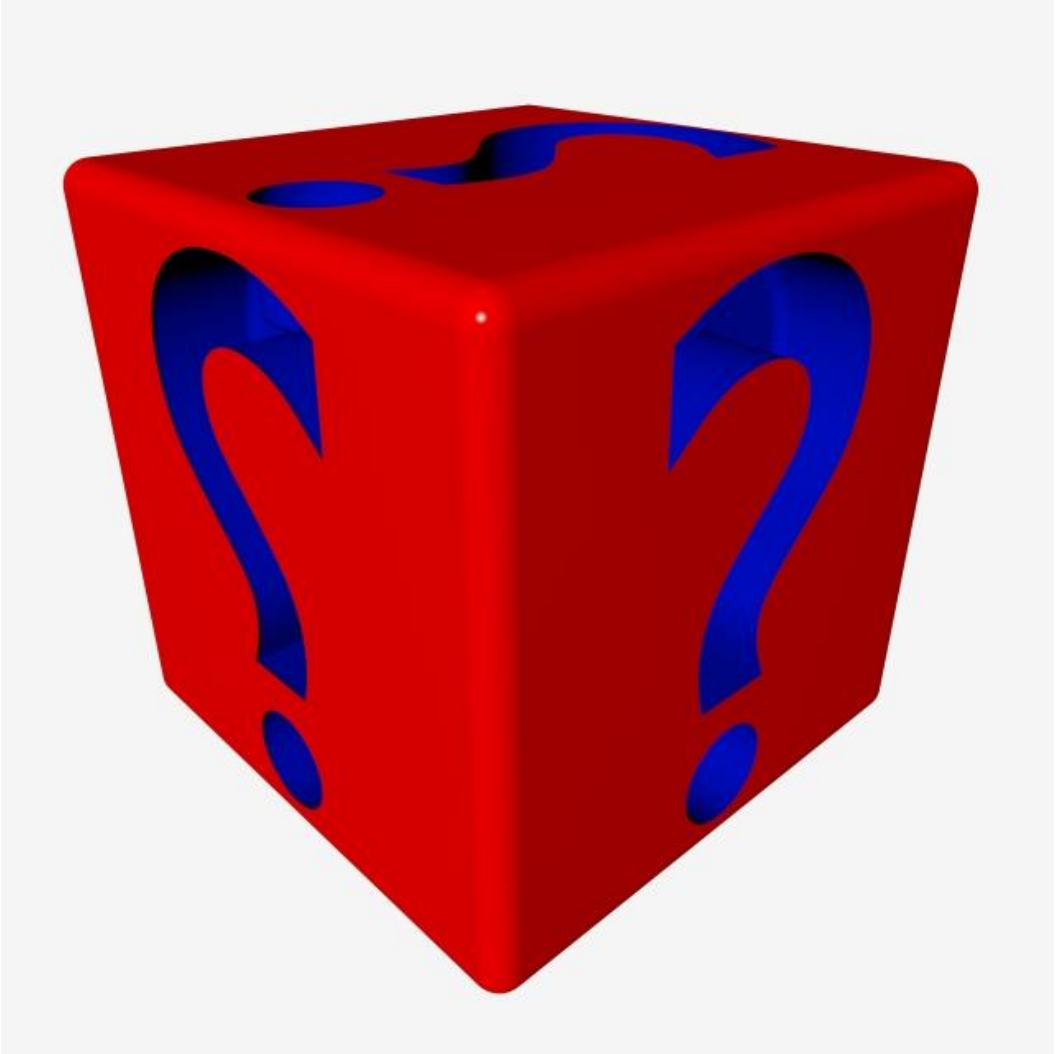
Ampicillina + gentamicina

UNA "TACHIPNEA SILENTE"

PEGGIORAMENTO CLINICO

FR 98 atti/min
Estremità fredde
Refill > 2"
EGA: pH 7.21; pCO2 27 mmHg; pO2 99 mmHg;I HCO3 12 mmol/L; BE – 17 mmol/L; lattati 11 mmol/L
Diuresi < 1 ml/Kg/h
T 35° C
PA arti superiori: 90/45 mmHg

UNA "TACHIPNEA SILENTE"

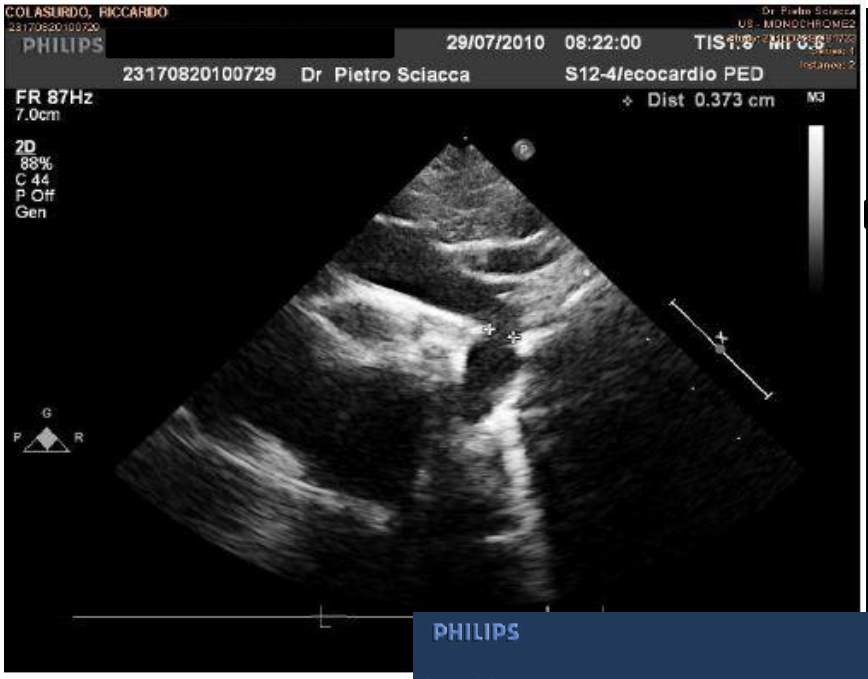


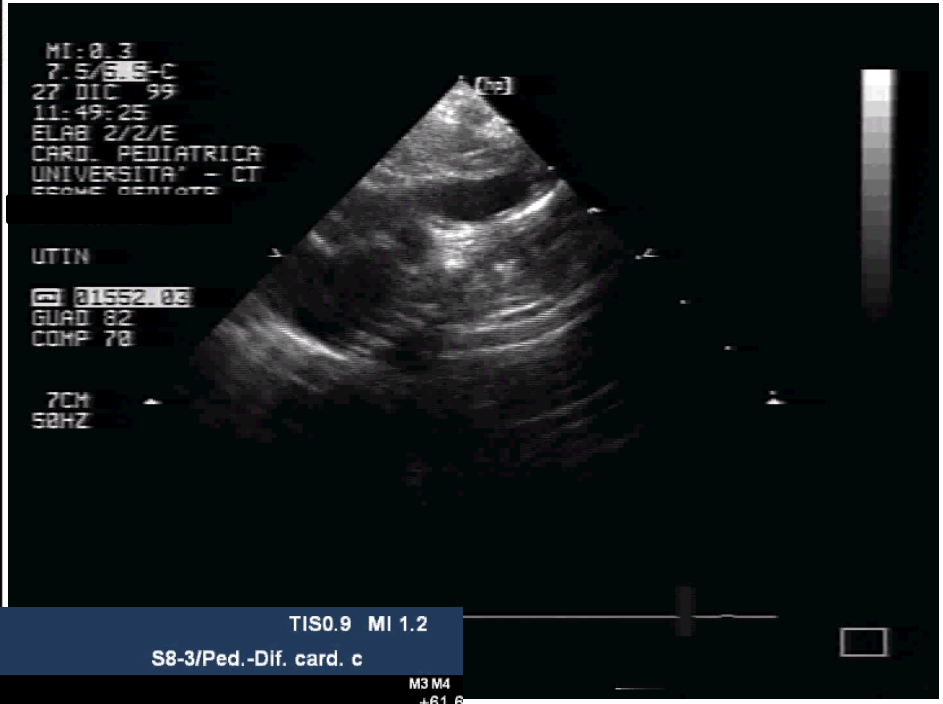
UNA "TACHIPNEA SILENTE"

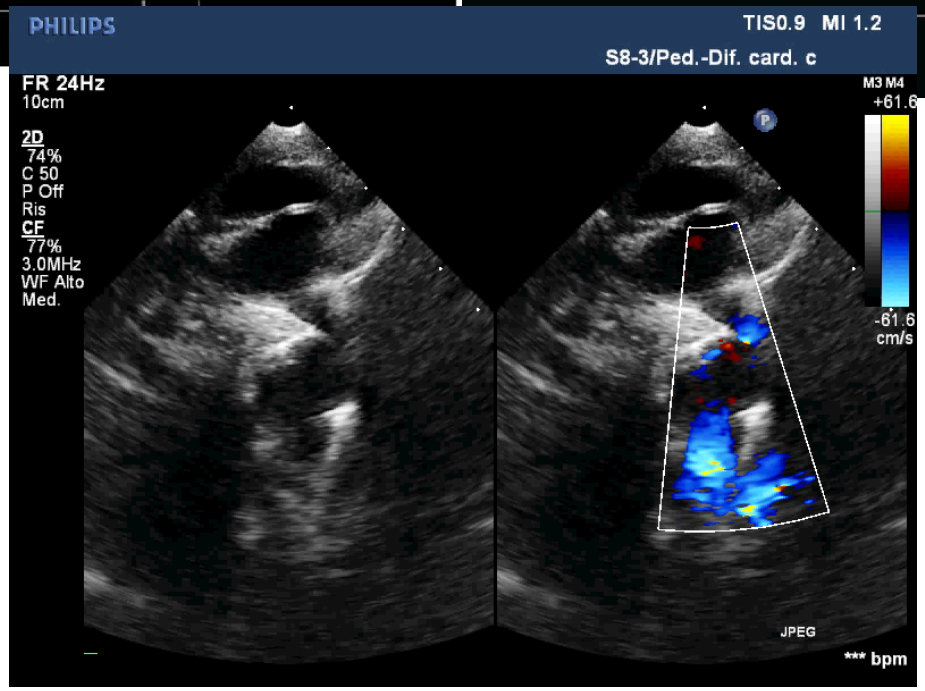
POLSI FEMORALI IPOSFIGMICI
PA arti inferiori non rilevabile
SpO2 arti inferiori< 4 % a SpO2 arti superiori

ECOCARDIOGRAMMA-COLOR DOPPLER

CoA


COLASURDO, RICCARDO
PHILIPS
29/07/2010 08:22:00
23170820100729 Dr Pietro Sciacca
S12-4/ecocardio PED
FR 87Hz
7.0cm
2D
88%
C 44
P Off
Gen
Dist 0.373 cm
M3


MI:0.3
7.5/6.5-C
27 DIC 99
11:49:25
ELAB 2/2/E
CARD. PEDIATRICA
UNIVERSITA' - CT
UTIN
31552.03
GUAD 82
COMP 70
7CM
50HZ


PHILIPS
TIS0.9 MI 1.2
S8-3/Ped.-Dif. card. c
FR 24Hz
10cm
2D
74%
C 50
P Off
Ris
CF
77%
3.0MHz
WF Alto
Med.
M3 M4
+61.6
-61.6
cm/s
JPEG
*** bpm

CONCLUSIONI

- Deficit di suzione: spesso primo segno clinico di emergenza neonatale

- Emergenze neonatali: spesso durante la prima settimana di vita

- Pediatra di famiglia: corretto iter diagnostico e diagnosi precoce

Ultima riflessione

A volte disordini estremamente rari possono mimare patologie comuni ma è necessario sospettare anche l'eventualità di patologie rare al fine di poter giungere alla diagnosi

Grazie !