

First International Conference on Prevention of FASD

Edmonton, Alberta / Canada, September 23-25, 2013

An welche Differenzialdiagnosen sollte gedacht werden,
bevor die Diagnose FASD gestellt wird?
(FASD = Fetal Alcohol Spectrum Disorders)
Auszug aus: FASD 2013 Epidemiology, kleanthes, 2013

Edmonton
Sept. 2013



Avoidance of false-positive findings

25 FASD differential diagnosis

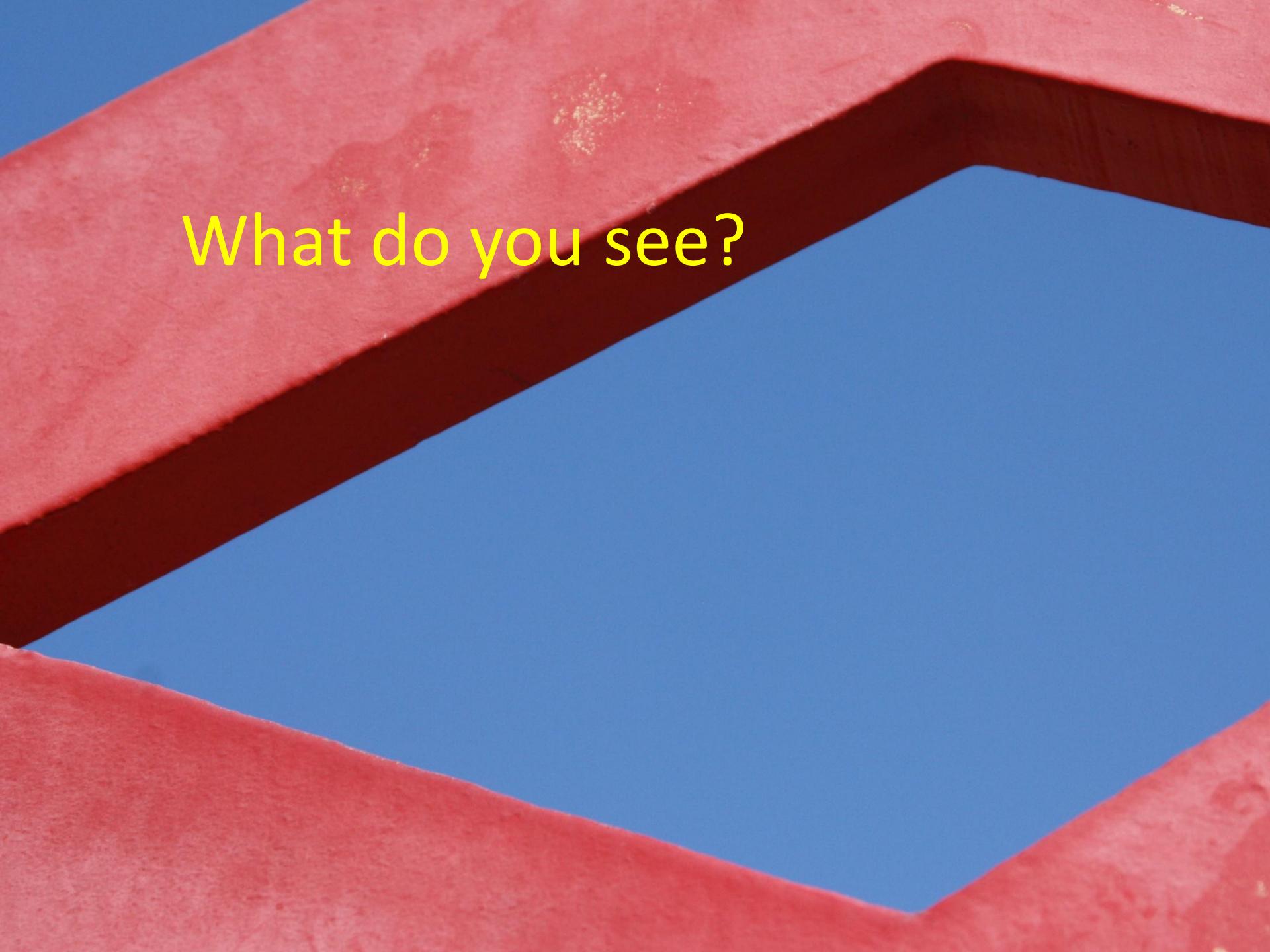
Differential diagnostics consist of „noticing them“
(Franz Volhard, 1872-1950)



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Edmonton, AB / Canada
1st Internat. Conference
on Prevention of FASD
September 23-25, 2013

Abstrakt book, p. 49-50; www.fasdedmonton2013.ca
www.kleanthes.de



What do you see?



That's the answer.

Avoidance of false-positive findings

25 FASD differential diagnosis

INTRODUCTION



To measure the effects
of prevention, clear-cut
diagnoses are required.

T H È S E
pour le
DOCTORAT EN MEDECINE

(*Diplôme d'Etat*)

par

ROUQUETTE Jacqueline

née le 19 Février 1925 à COURBEVOIE (Seine)

Présentée et soutenue publiquement le :

INFLUENCE DE LA TOXICOMANIE ALCOOLIQUE PARENTALE

SUR LE DÉVELOPPEMENT PHYSIQUE & PSYCHIQUE DES JEUNES ENFANTS

Président de Thèse : M. JULIEN-MARIE (Professeur)



Jacqueline Roquette (Paris, 1957):

Influence de la toxicomanie alcoolique parentale sur le développement physique & psychique des jeunes enfants

in:

FASD 2013 Epidemiology (EPUB)
kleanthes, Dresden 2013
Paditz E, Rissmann A, Götz D, Fitzpatrick P

**8.75 % (7/80) of cases
with „FASD“: another syndrome**

Douzgou, S. et al.:

Diagnosing fetal alcohol syndrome: new insights from genetic technologies.
Arch. Dis. Child. 2012: 97(9), 812-817

14 % false positives

psychological findings between FASD
and healthy subjects

Nash, K. et al.:

A differential approach for examining the behavioural phenotype
of fetal alcohol spectrum disorders.

J. Popul. Ther. Clin. Pharmacol. 2011: 18(3), e440-453

Avoidance of false-positive findings

25 FASD differential diagnosis

QUESTION



Avoidance of false-positive findings

25 FASD differential diagnosis

METHODS



Methods/Search:

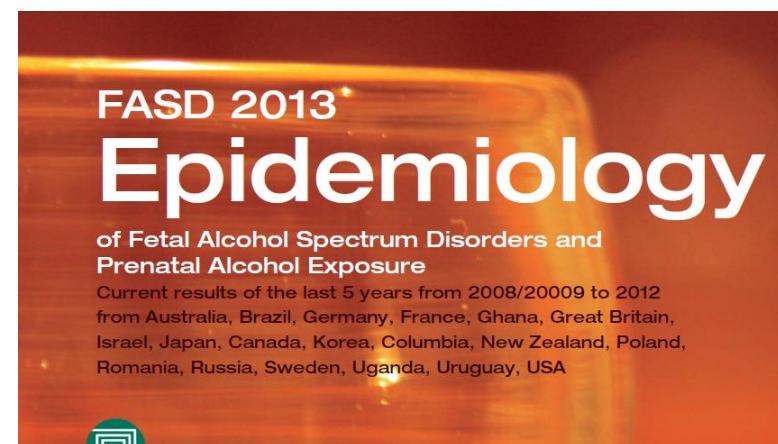
- textbooks
- review articles
- PubMed (morphological and psychological symptoms associated with FASD)

in: **FASD 2013 Epidemiology (EPUB)**

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Paditz E, Rissmann A, Götz D, Fitzpatrick P

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RESULTS



FASD Differential Diagnosis (I)

Syphilis connata

Fanconi anemia [49]

Dubowitz syndrome [52–56]

Williams syndrome

Noonan syndrome [57]

Silver Russell syndrome

Brachman-de Lange syndr.

Smith-Lemli-Opitz syndr. [49]

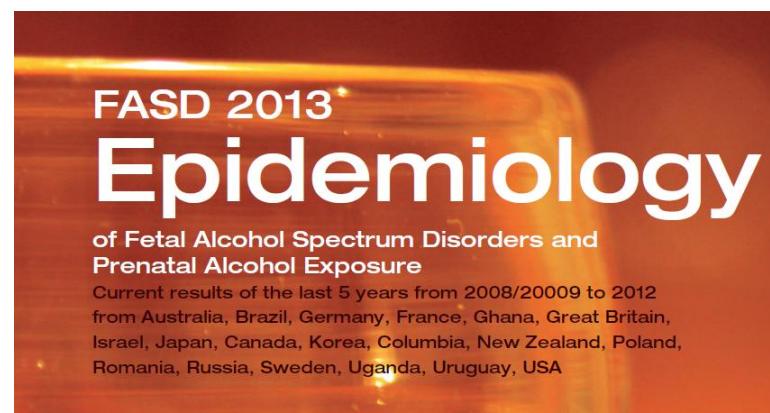
Stickler syndrome [49]

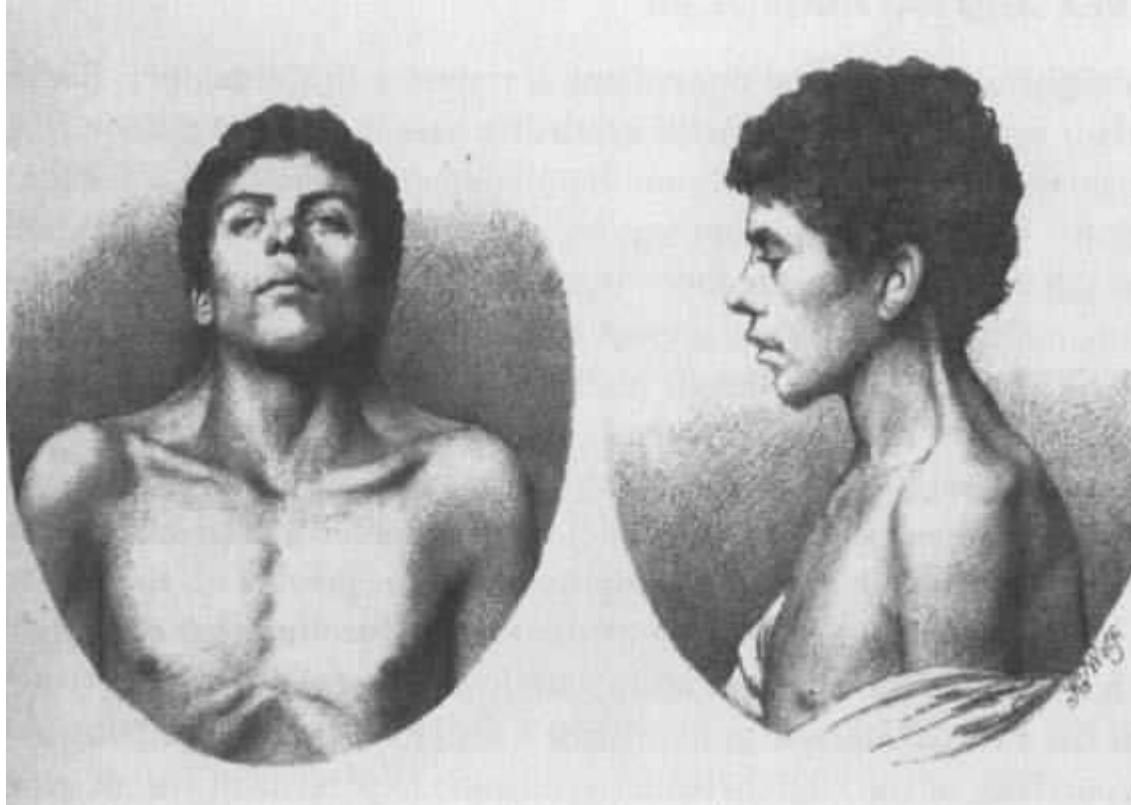
Turner syndrome [49]

Kabuki syndrome [49]

DiGeorge syndrome [58–59]

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Kobylinski O. :
Über eine flughaut-
ähnliche Ausbreitung
am Halse.
Arch Anthropol, 14, 343-348,
1883.

Noonan syndrome

The oldest known case of Noonan syndrome, described in 1883 by Kobylinski

Source: <http://en.wikipedia.org/wiki/File:Noonan1883.JPG>

FASD Differential Diagnosis (II)

SMADIP1-Mutationen [60]

22q11 abnormalities, e.g. 22q11 dup, 22q11 del [49]

Micro-deletions und Micro-duplications,

e.g. 1q21 dup, 2p16.1p15 del, 14q21.1 dup / 16p13.3 dup, 15q13.3 del [49]

fetal Valproate syndrome

fetal Hydantoin syndrome

Toluene embryopathy [61]

Autism [51]

Reactive attachment disorder [51]

ODD or conductant disorder [51]

Pervasive developmental delay [51]

Learning disabilities [51]

Emotional / behavior problems [51]

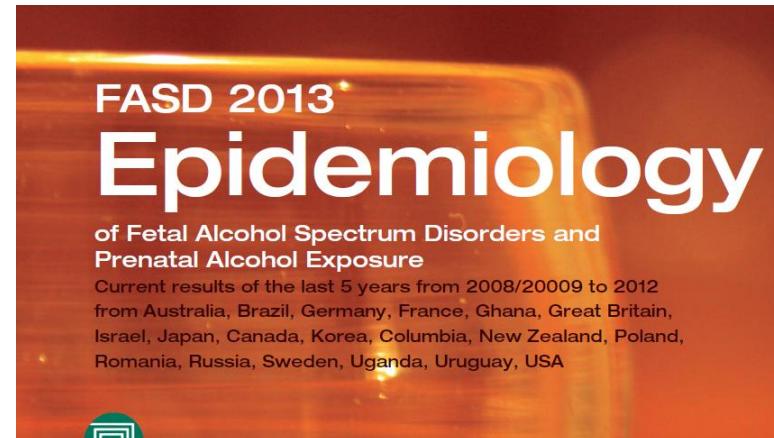
ADHD [51]

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CONCLUSIONS





Differential diagnostics consist of
„noticing them“
(Franz Volhard, 1872-1950)

The overlapping of morphological findings between FASD and other syndromes as well as the similarity of psychological findings in children with FASD, ADHD and other disorders, poses therefore in practice, a significant challenge that can only be mastered interdisciplinary.

THANK YOU



Edmonton
September 22, 2013

