1° Central & Eastern European Summit on Preconception Health and Prevention of Birth Defects

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Key Issues to Have a Good Birth Defects Surveillance and Research Program in the Early 2000s

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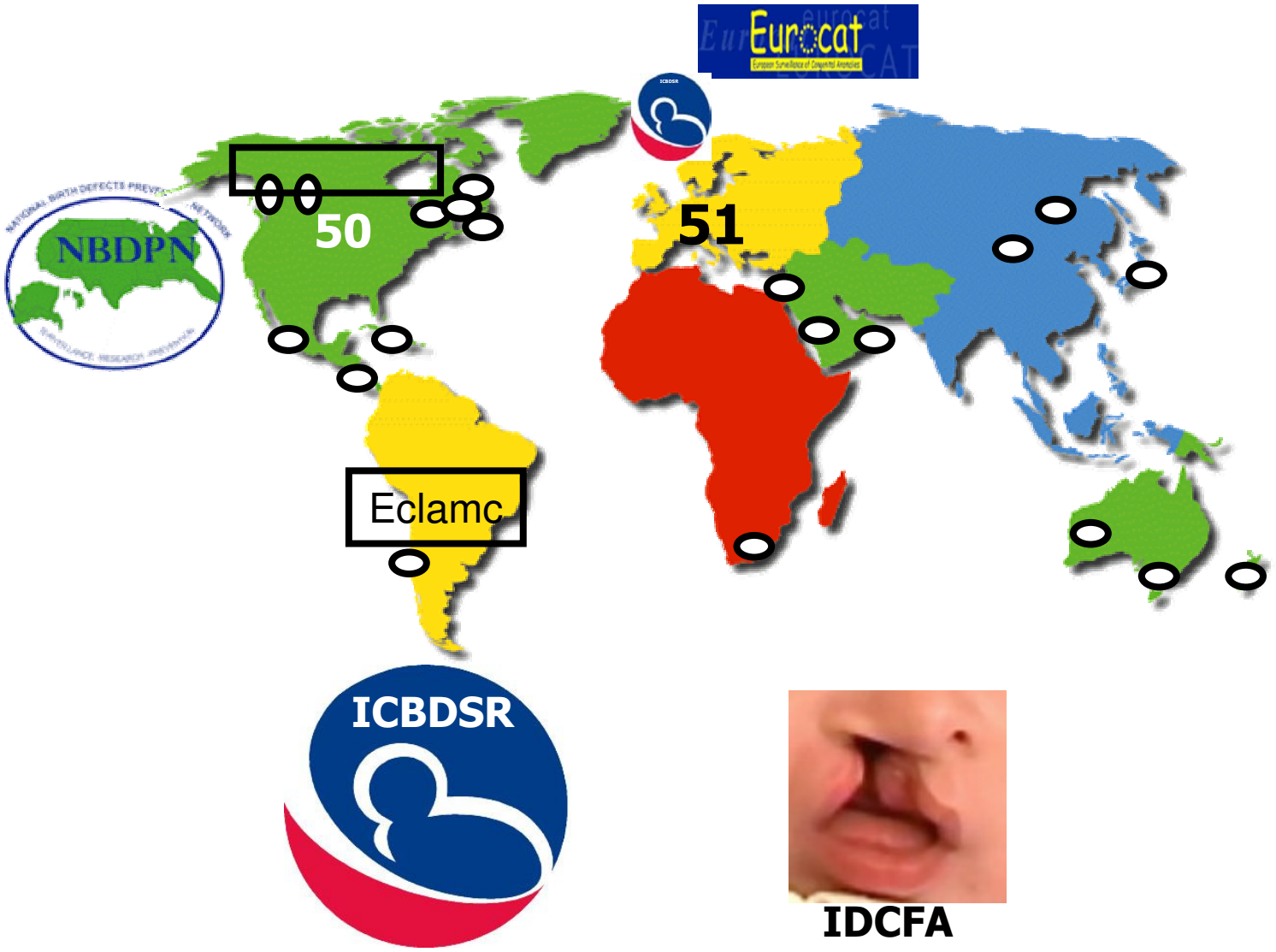
Summary

- I will examine the main and common objectives planned by the existing Birth Defects Surveillance and Research Programs (BDSRP) and their Networks

- I will try to discuss if all BDSRP have achieved the planned objectives and I will speculate on how to improve the achievements

- I will stress that a new paradigm is necessary for all BDSRP to be implemented in the next few years

123 BDSRP, 3 Networks



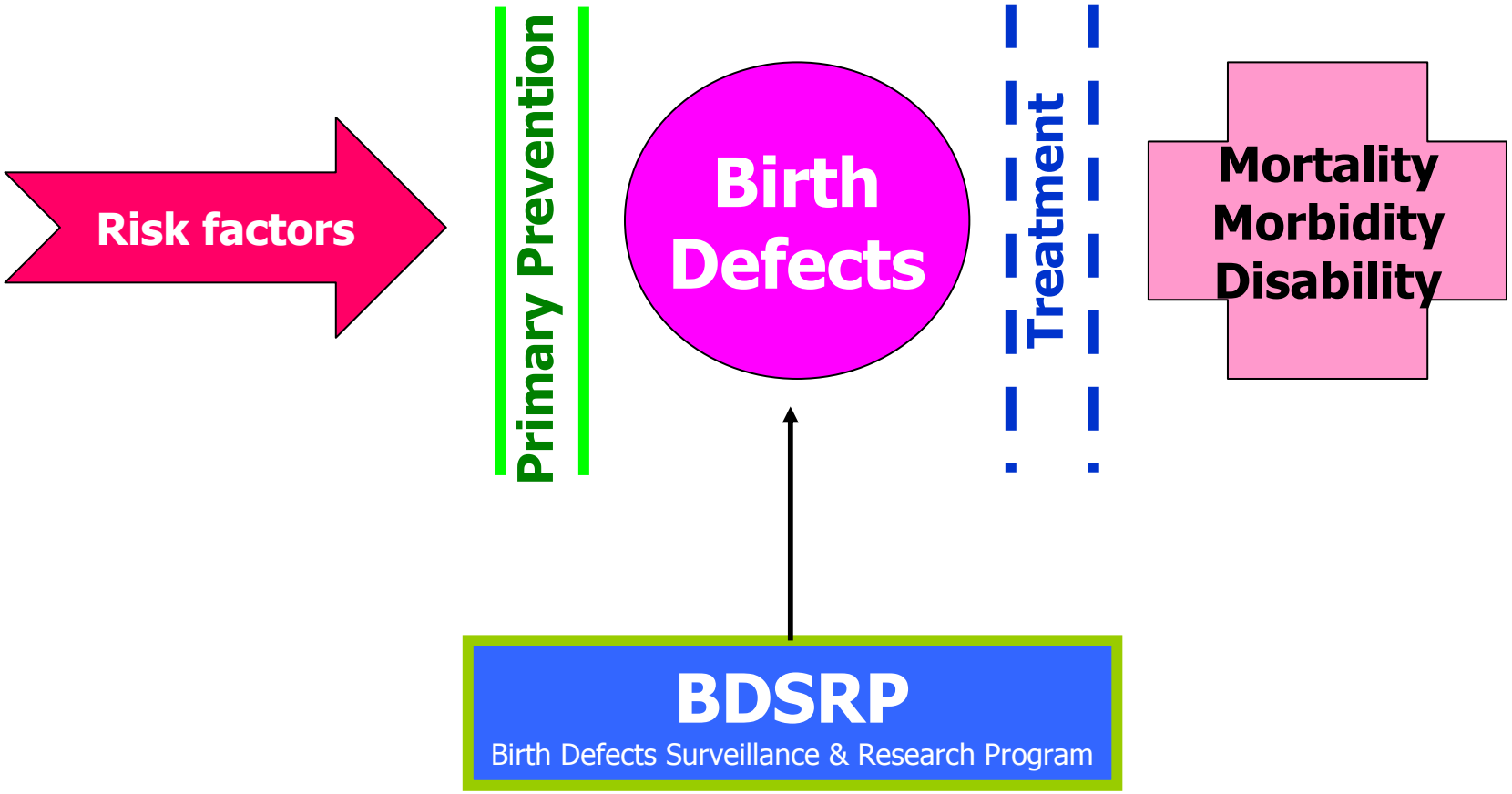
IDCFA

BDSRP and Networks Common Aims

1. Providing and exchanging epidemiologic information on Birth Defects
   - Descriptive epidemiology, including quantifying morbity and mortality, evaluating time trends and population variations
2. Using one or more early warning methods to detect possible new teratogenic exposures
   - Birth Defects frequency monitoring, cluster identification, unusual cases and associations
3. Conducting or facilitating researches related to the causes and prevention of Birth Defects
4. Evaluating the effectiveness of prevention

5. *Providing information for community concerns, education advocacy*

Chain of events related to birth defects and the focus of BDSRP



The two main questions

- Have the main objectives been achieved by all the BDSRP ?

- How can we improve the achievement of the objectives ?

Aim # 1

Provide and exchange epidemiologic information on birth defects

- All BDSRP provide and exchange epi information on birth defects

- However:
  - The format for presenting data is often unclear or insufficient
  - Not all BDSRP produce and disseminate a full periodic report and/or have a fully informative website
  - Most of the time data are related only to “structural” birth defects *(Congenital malformations,deformations and chromosomal abnormalities, ICD X° Chapter XVII),* other birth defects and other adverse reproductive outcomes (ARO) are usually excluded
  - The validity of registered cases and related information is rarely evaluated

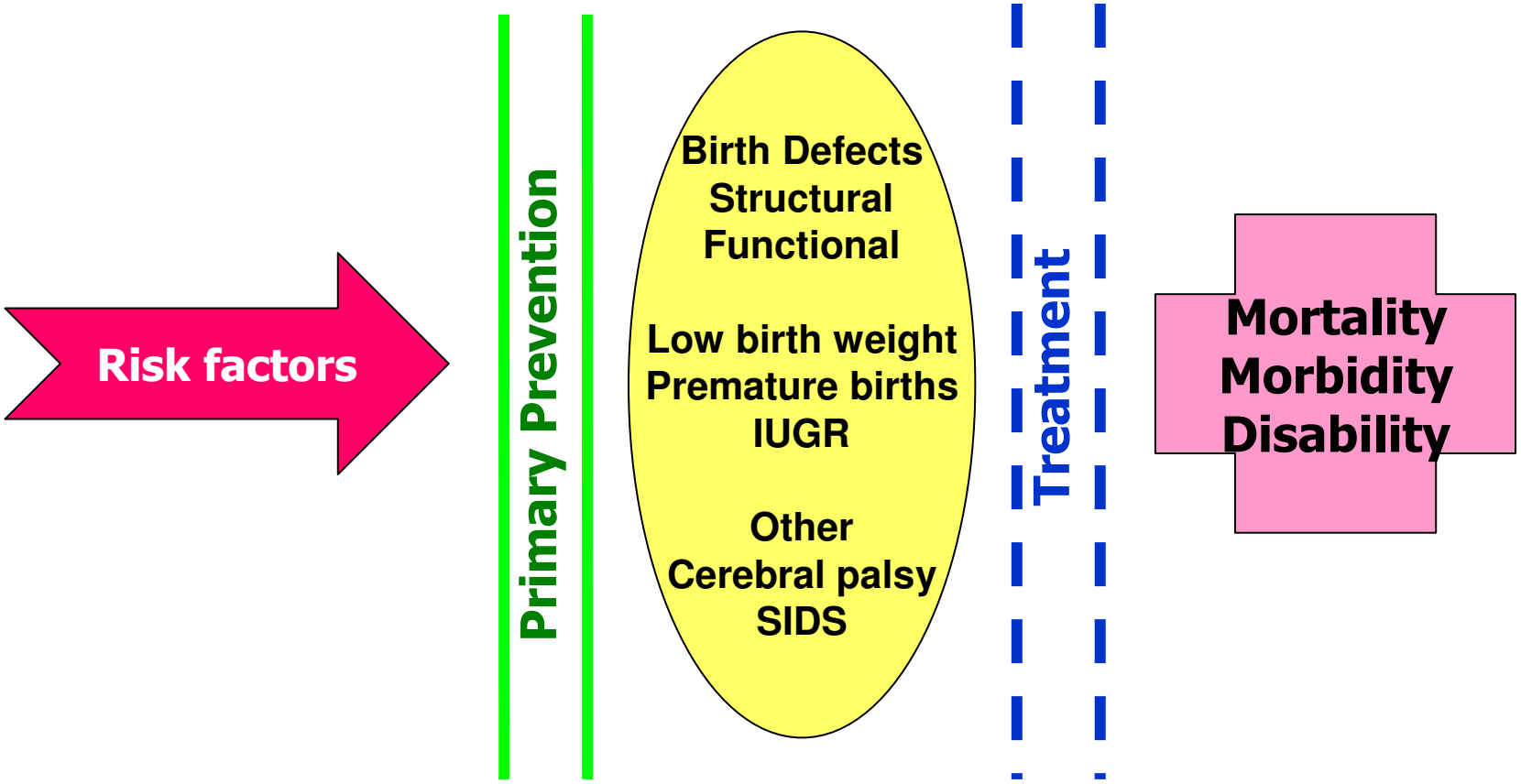
Aim # 1

Provide and exchange epidemiologic information on birth defects

- All BDSRP provide and exchange epi information on birth defects

- However:
  - **Almost always data are related only to "structural" birth defects *(Congenital malformations,deformations and chromosomal abnormalities, ICD X° Chapter XVII),* other birth defects and other adverse reproductive outcomes (ARO) are usually excluded**

Chain of events related to BD & other ARO from the perspective of primary prevention and preconception care



Aim # 1

Provide and exchange epidemiologic information on birth defects

- All BDSRP provide and exchange epi information on birth defects

- However:
  - **The validity of registered cases and related information is rarely commented and evaluated**

Reported total prevalence (x 10,000) of Esophageal atresia from 41 BDSRP *(ICBDSR data, 2001-2005)*

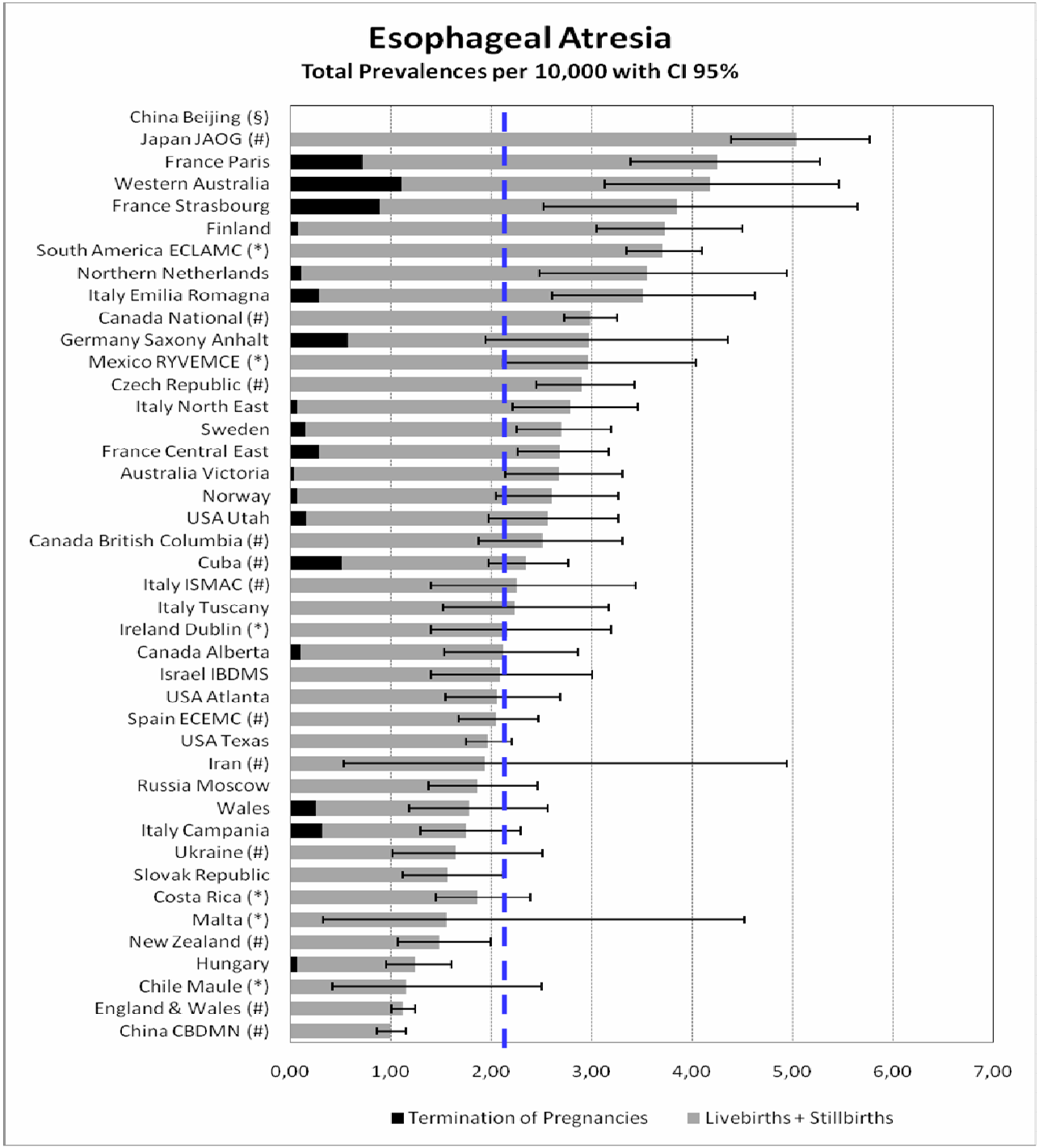
How to be confident to the data for :

- Promoting research
- Evaluating preventive measures
- Quantifying morbity and mortality
- Planning services

Without a validation

?

(§) Defect not reported
(*) ToPs not permitted
(#) ToPs not registered (or partially registered) although permitted



Aim # 2

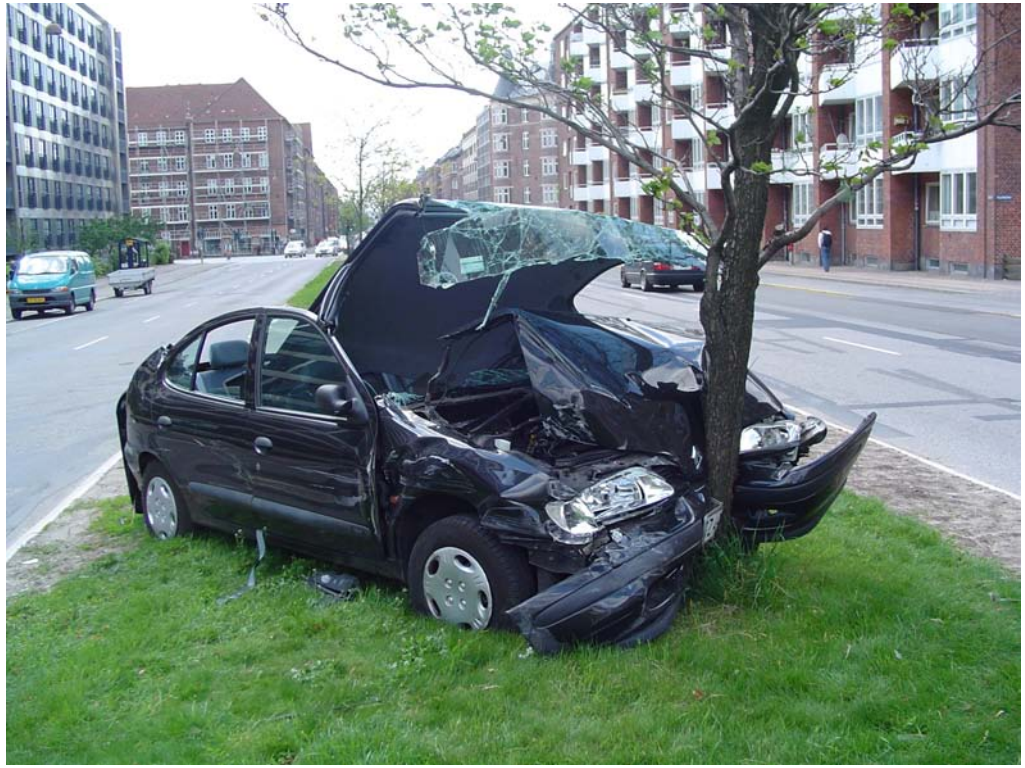
Facilitate the early warning of new teratogenic exposures

- The early warning (monitoring of prevalence of birth defects, more or less specific) is systematically performed in a few BDSRP.
- However :
  - When done, is late and far from to be specific as needed
- Although, as a matter of fact:
  - an early warning of new teratogenic exposure is never been provided by any BDSRP
  - in the last decades a few “new” teratogenic exposures have been discovered (the negative results may be true !)

Aim # 2

Facilitate the early warning of new teratogenic exposures

- The early warning activity is like the “insurance against car accidents”
- **It must be done**
- It must be done properly although no accident happened for years



Aim # 3

Conduct or facilitate the research related to the causes and prevention of birth defects

- Up to now one of the most successfull achievements obtained by some BDSRP

- However:
  - Mainly by Atlanta and California Programs, and more recently by the US NBDPN Network
  - Exceptions in other countries: Hungary, and partially others (e.g.: Norway, France CE, Italy IPIMC)

- As a matter of fact:
  - Human resources and funds are lacking in many programs
  - International collaborative studies have been rarely performed
  - New study methodologies must be expanded (e.g.: genome wide scan, gene-environment interaction)

Aim # 4

Evaluate the effectiveness of prevention of birth defects

- It is a clear “direct product” of surveillance systems (descriptive epi, used in ecological correlation studies)
- Performed by many BDSRP

- Best examples
  - Recommendations for folic acid supplementation has no (not yet ?) impact on population prevalence of NTD or other malformations (Eurocat and ICBDSR studies)
  - Folic acid fortification reduces the population risk of NTD and possibly of other defects (Many studies)
  - Moreover, good evidence from many BDSRP on the *birth prevalence* decrease of many birth defects linked to prenatal diagnosis and ToP

- However
  - Few studies on effectiveness of preventing of other risk factors (e.g.: AEDs, diabetes, smoking, alcohol)

Aim # 5

Provide information for community concerns, education, advocacy

- No data

- However, together with other organizations (e.g.: TIS, non-profit associations)
  - attention to the problems related to birth defects has increased
  - community concerns have been well addressed
  - education and advocacy have been mantained

Answer # 1

Have the main objectives been achieved by all the BDSRP ?

- The main aims have been achieved by few BDSRP, only partially by others
- Great differences between the various BDSRP are clear
- The great differences between the various BDSRP are related to the human resources and funding available to each program

Answer # 2

How we can improve the objectives achievements ?

Since public health policy makers are obviously attracted by concrete results …

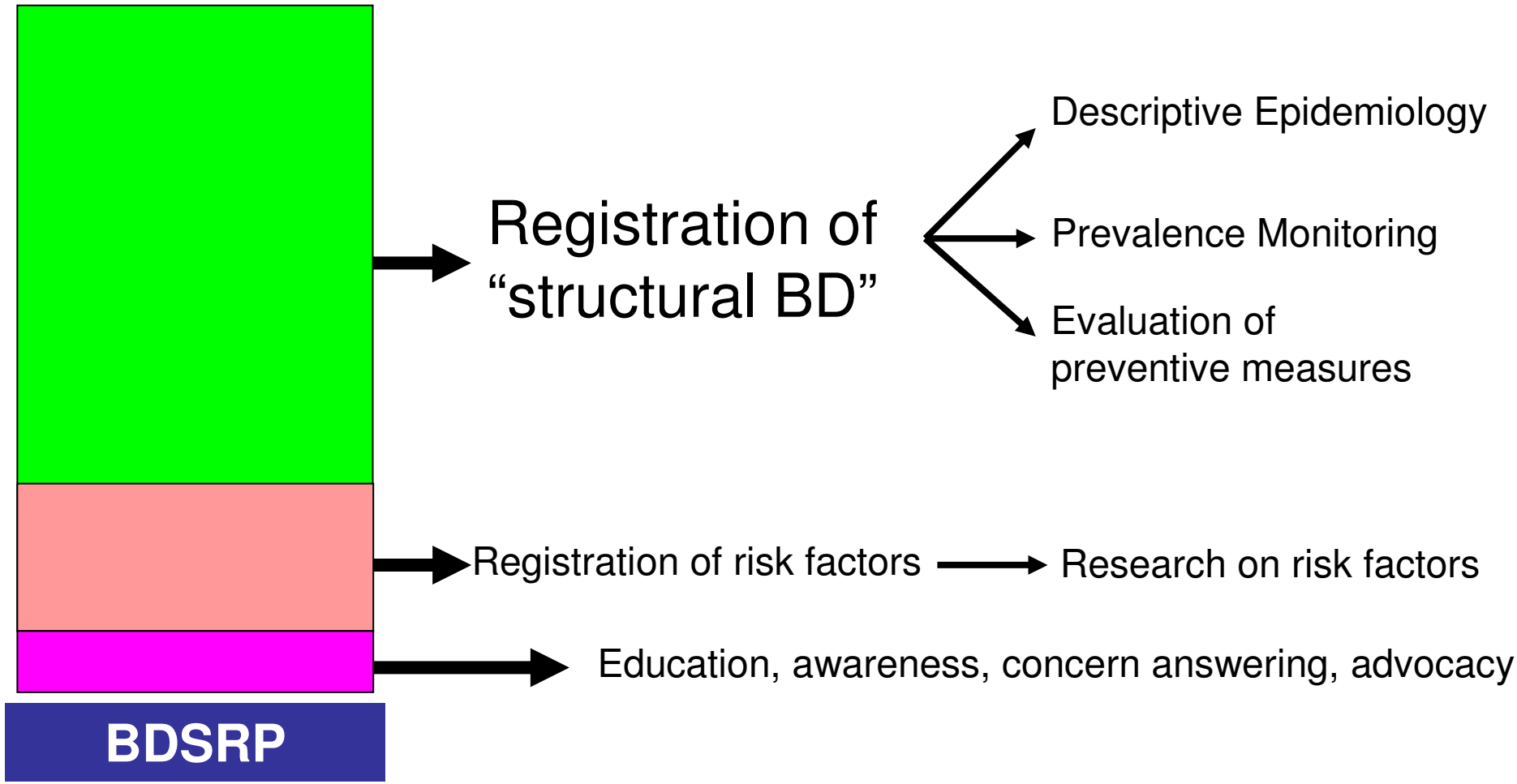
… more human resources and funding can be obtained if …

… BDSRP adopt a new paradigm: **“mother & child - oriented”**

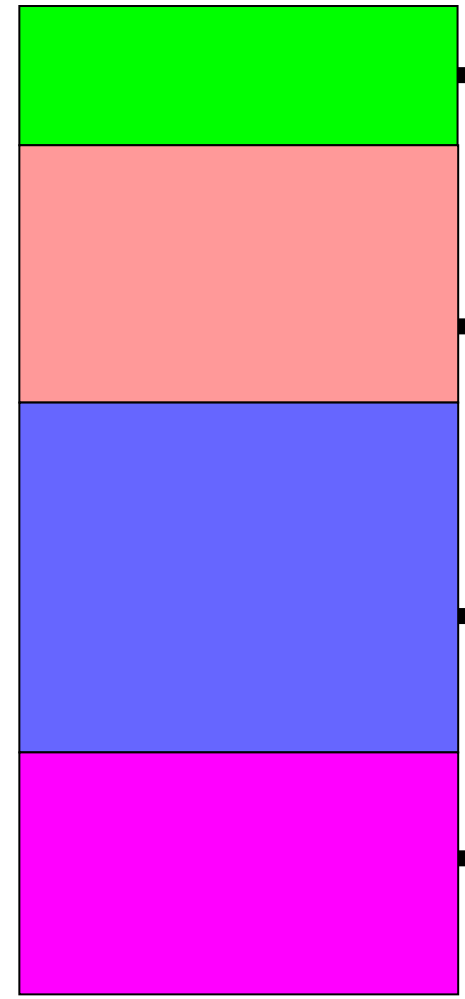
The old paradigm: birth defects - oriented

- Most of the BDSRP have been in fact:
  - implemented, even decades ago as an answer to :
    - the thalidomide tragedy (e.g.: England & Wales, Sweden, CDC-Atlanta),
    - other public concerns (e.g.: in Italy 1976 Seveso, New York 1978 Love Canal, Texas 1993 cluster of NTD)
  - planned for:
    - quantifying the frequency in the target population of birth defects
    - an early detection of a new possible epidemic of a birth defect
    - evaluating the role of the environmental pollution (e.g.: small areas studies, time trends)

The old paradigm: birth defects-oriented



The new paradigm: mother & child - oriented



Registration of all ARO & BD

- Descriptive Epi including evaluation of preventive measures
- Outomes Prevalence Monitoring
- **Validity studies**

Registration of risk factors Env + mat + gen

- Risk Factors Prevalence Monitoring
- Research on Risk Factors
  - Gene-environment
  - International

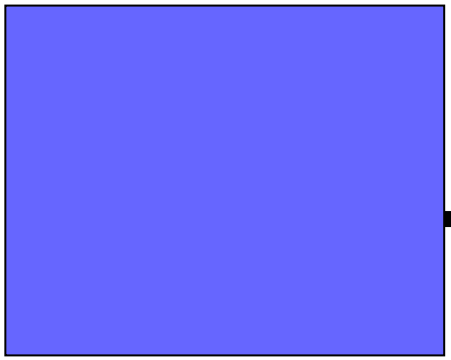
Registration of Health status of M & C Services answers

→ **Evaluation**
QoL, QoS, Cost

Education, awareness, concern answering, advocacy

BDSRP

The new paradigm: mother & child-oriented

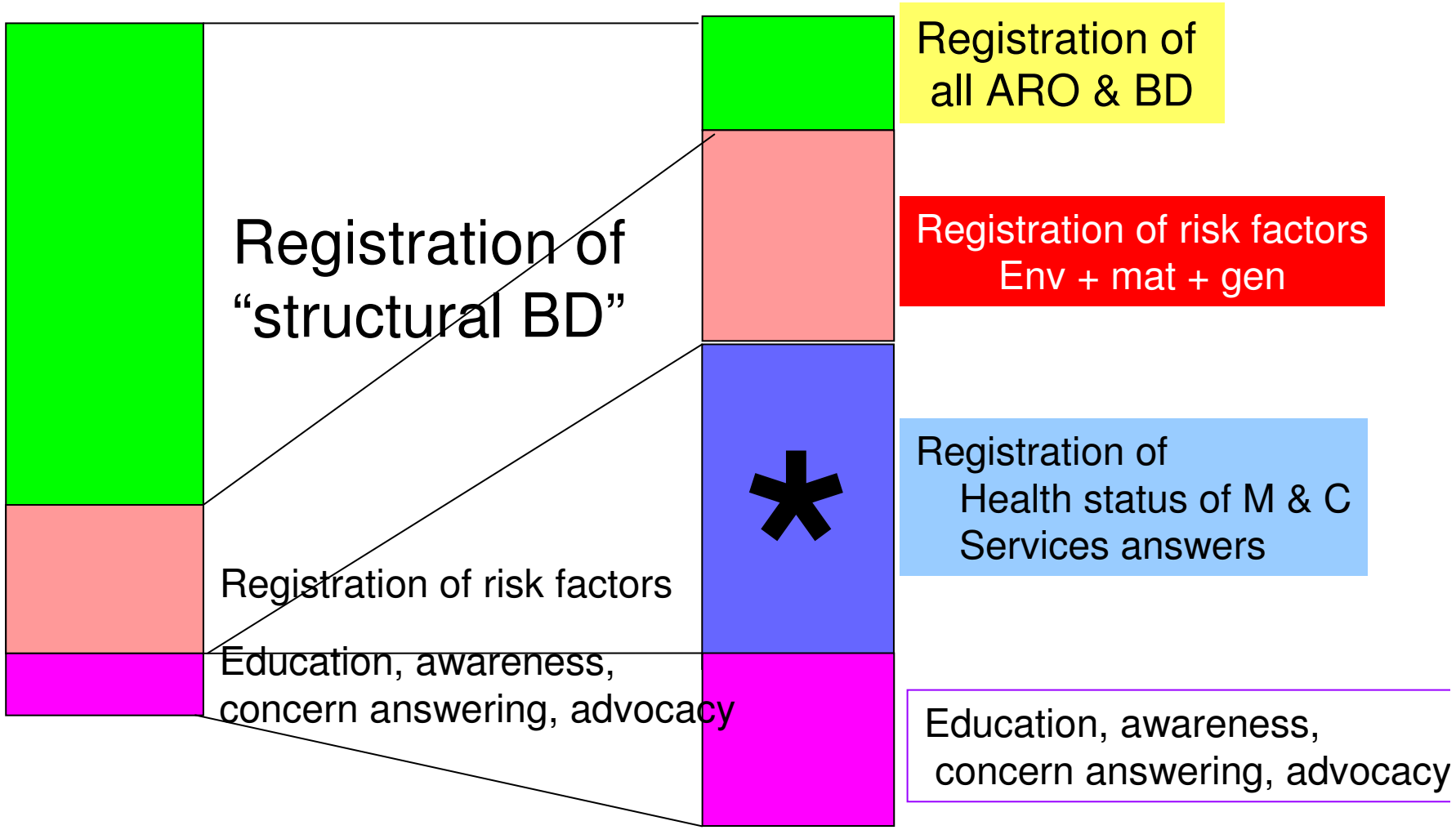


Registration of
- Health status of mothers and children
- Preconception, prenatal and postnatal services answers

Evaluation
QoL, QoS, Cost

BDSRP

Look at the different proportion of the main activities



Some key issues

- Use your infrastructure and knowledge for re-styling the program in a population-based surveillance and research of all adverse reproductive outcomes and birth defects (ARO & BD)
  - The so called “opt-in” should be fought and refused since it distorts any result and may give false messages. Data protection and security can be obtained with other approaches.

- Try to have the most valid data (active ascertainment, multiple sources, linkage with other databases)
  - Outcomes
    - Internal and external population differences, time trend, monitoring
  - Risk factors
    - Nutrition, nutritional supplements, medications, smoking, alcohol, maternal diseases, environmental characteristics

Some key issues

- Classify ARO & BD involving field clinician experts
  - Phenotype characterization to define more etiologically homogeneous categories is essential.
  - A clinician is essential in coding
- Give to the lay and scientific community fully informative tables and figures and the evidence of the validity of results
- Promote or collaborate to
  - Studies on gene-environment interaction
  - Studies on health of children with ARO & BD, registering:
    - Timing (pre-postnatal) of diagnosis
    - QoL & QoS parameters
    - Cost of medical care
  - Education, awareness,advocacy
    - Social marketing campaign targeting smoking, alcohol, risky medications, preconception care

Conclusion

- Congratulations to the organizers of this conference

Look here !!!

Conclusion

- As you can see the “Europe” boat appeared and could not start from a better city

Conclusion

The lectures of this conference clearly underscore that the ultimate scope of a modern surveillance program is actually to provide support to the preventive and treatment services

to alleviate the tremendous burden related to the many risk factors acting in the first week of prenatal life of the human beings

preventable only improving the women health and the preconception services