

Very Rare Disorders *organisation of care*

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Rare Disorders

- <1:2,000

Example: Marfan syndrome

- 1:10,000

- Number in Europe 75,000

Very Rare Disorders

- <1:2,000,000
- Numbers in Europe 50-300/entity

Examples

del11qter

Progeria



Very Rare Disorders

Example

- Marshall-Smith syndrome
- Number in Europe 30
- Sporadic
- No cause known

Ultra Rare Disorders

- <1:100,000,000
- Numbers in Europe 5-10

Ultra Rare Disorders

Example: Causes of mental retardation

- Diagnosis in 50-55%
- Genetically determined 75-80%
- Three most common

Down syndrome

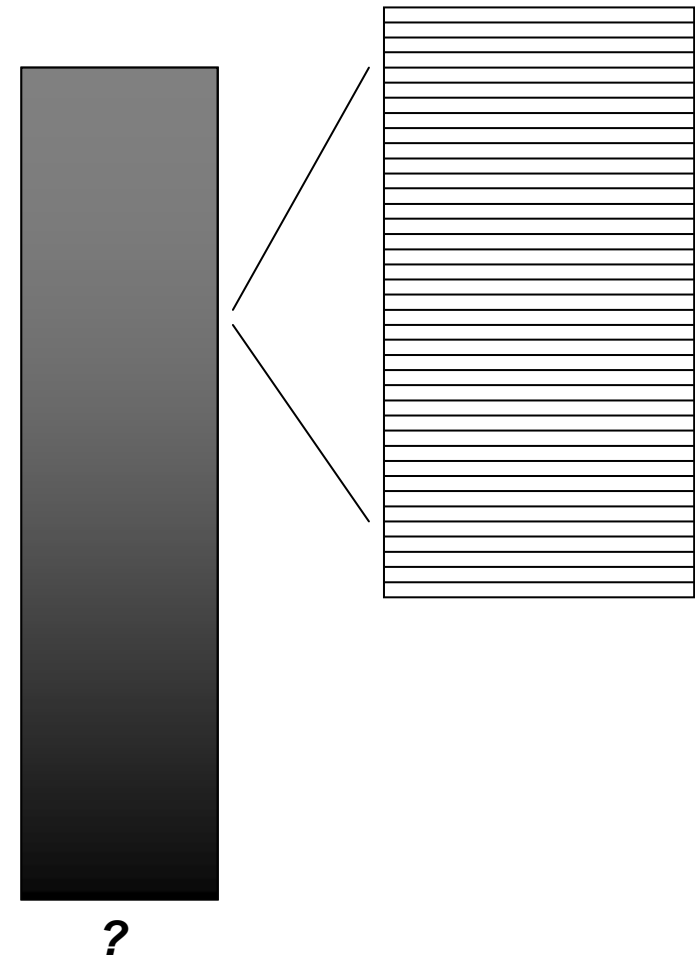
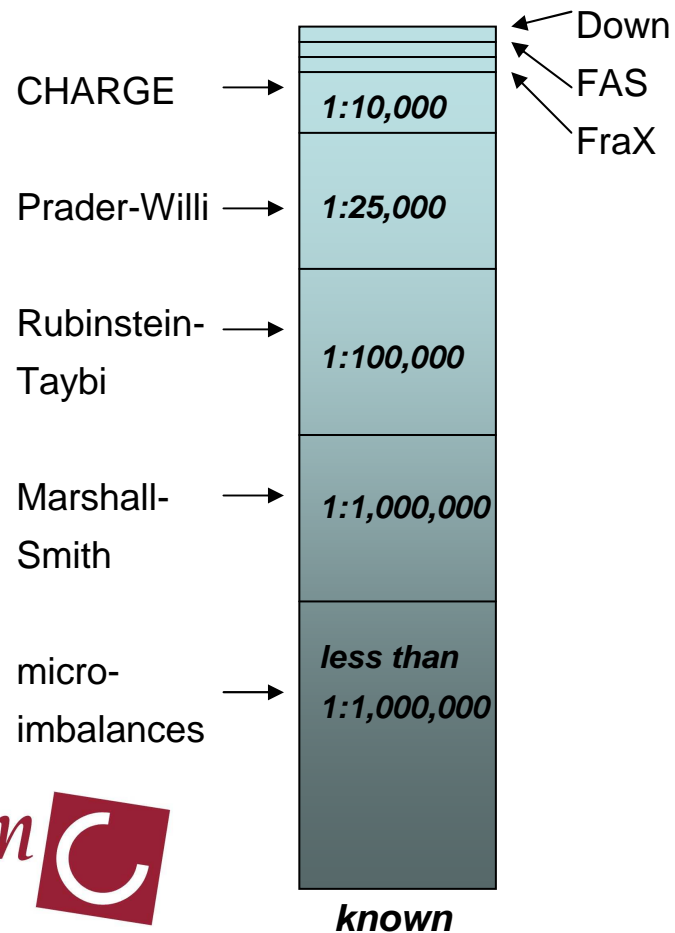
Fetal Alcohol

Fragile X



Ultra Rare Disorders

Example: Causes of mental retardation



Ultra Rare Disorders

Example: Causes of mental retardation

- Diagnosis in 50-55%
 - Genetically determined 75-80%
 - As yet unknown group 35-40% genetic cause
 - Prevalences 1:10,000,000-100,000,000
- *total number in Europe 5,000,000-10,000,000*

Ultra Rare Disorders

Medical care

- Not fragmented
 - Reachable
 - Affordable families
 - Affordable countries
- *centres for rare diseases*
1/country or 1/30,000,000 inhab

Ultra Rare Disorders

Medical care

Issues

- Languages
- Costs hospital
- Costs stay
- Transportation information

→ *prerequisites:* *Communication*
 Trust



Ultra Rare Disorders

Medical care

Further Issues

- Choice disorder / center
- Grouping
- Choice locations
- Flagging
- Research



Ultra Rare Disorders

Medical care

Example Marshall-Smith

- One hospital
- E112 form
- Ronald McDonald house
- Flagging though support group
- e-mail consultations
- Research



Ultra Rare Disorders

‘Mutual’ care

- Europe-wide Registration
- Support groups
- Site
- e-mail consultations

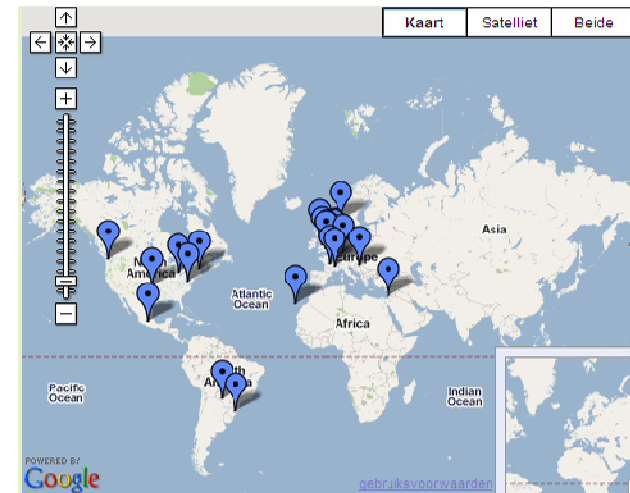


Ultra Rare Disorders

‘Mutual’ care

Example Marshall-Smith

- Active parents
- Site



Ultra Rare Disorders

‘Mutual’ care

Example Marshall-Smith

- Active parents
- Site
- Wiki

Home > Research > MSS wiki > Marshall-Smith Syndrome (introduction)

- [Login](#)
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MSS TRANSLATION

English

GOOGLE TRANSLATE

Selecteer een taal

Mogelijk gemaakt door Vertalen Google

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Marshall-Smith Syndrome (introduction)

 In 1971, Drs RE Marshall, CB Graham, CR Scott, and DW Smith reported two unrelated male children with unusual facial features, poor weight gain, delayed development, and what appeared to be advanced maturation of the bones. The faces of the two children looked similar in that they had prominent eyes and thick eyebrows. The top of the nose protruded less than normal and the nose was small with an upturned tip. Both children had breathing problems and one of them died at 20 months of age.

Since then, approximately 60 children have been reported in the medical literature, and the condition has become known as Marshall-Smith syndrome (MSS). Most of the children described in the early medical reports died at a young age, but when treatment of the breathing difficulties improved many children survived, although there is still early mortality.

Here we provide an update of the medical condition of four of the children previously described in the medical literature, and describe the medical features of ... new children we have seen. Three of the patients are aged over 16 years, and therefore provide hitherto unknown information of MSS in adulthood.

[> MSS wiki](#)

Ultra Rare Disorders

'Mutual' care

Issues Marshall-Smith

- Same as for Medical care

Ultra Rare Disorders

Suggestion

- In FP8 specific call for centers for rare disorders
- Facilities local governments
- Costs insurance companies