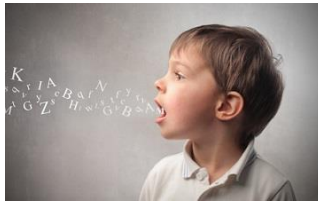


## Assessment of language development in children

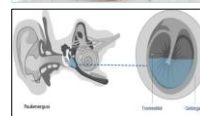


Jochen Rosenfeld  
Abt. Gehör-, Sprach- und Stimmheilkunde  
Kinderaudiologie und Logopädie

Kantonsspital St.Gallen – ein Unternehmen, drei Spitäler. St.Gallen Rorschach Flawil

## What does language have to do with medicine?

- Developmental disorders of speech and language (secondary)
- Indication for a surgical treatment (otitis media with effusion)
- Monitoring success of hearing aids
- Early detection – secondary prevention



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## Overview


I.	Starter: Clinical case
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## Clinical case

Jonas, 3;3 years old  
Developmental disorder of speech and language


### Questions

- What is Jonas telling us?
- What do you think about his language comprehension?
- What do you think about his language production?
- What are his compensation strategies?

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# Clinical case

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## Language development

- Comprehension



- Production



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## Linguistic levels

- Phonetics – Phonology
- Semantics – Lexicon
- Morphology – Syntax
- Pragmatics – Communication



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## Classification

- Environmentally dependent language abnormalities (Umgebungsbedingte Sprachauffälligkeit)
- Language delay (Sprachentwicklungs-Verzögerung)
- Language disorder (Sprachentwicklungs-Störung)

Interdisziplinäre Leitlinie Diagnostik von Sprachentwicklungsstörungen 2012  
<http://www.awmf.org/leitlinien/detail/II/049-006.html>


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## Environmental language abnormalities

- Lack of language stimulation/  
insufficient language influence
- Bilingualism / multilingualism




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## Language Delay

- up to 36 months
- «Late Talker»
  - from 24 months of age
  - < 50 words
  - no word combinations




```

            graph LR
            A[«Late Talker»] --> B[«Late Bloomer»]
            A --> C[Language disorder]
            
```

Age (years)	0.	1.	2.	3.	4.	5.	6.	7.	....
Diagnosis	Language Delay			Language Disorder					

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## Language disorder

- Temporal and content deviation of normal patterns of language acquisition
- Language comprehension (receptive) and language production (expressive)
- Linguistic levels (e.g. articulation, vocabulary, grammar, sentence structure)

Interdisziplinäre Leitlinie Diagnostik von Sprachentwicklungsstörungen 2012  
<http://www.awmf.org/leitlinien/detail/II/049-006.html>

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## Language disorders (Classification)

As a consequence of other diseases (secondary)

- Hearing impairment
- Mental retardation
- Autism
- Neurological abnormalities
- Syndrome



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## Language disorders (Classification)

Primary (isolated) language disorder

- Specific language impairment (SLI)
- Specific developmental disorder of speech and language (ICD: F80.1; F80.2)
- (Familial language weakness = Familiärer Sprachschwächetypus)
- (Developmental dysphasia = Entwicklungsdysphasie)

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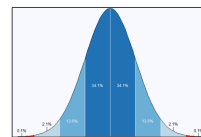
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## Language development

- Normal language acquisition
- Diagnostic cut-off between normal and impaired language development
- Possibilities of language assessment in clinical practice



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## Traits, skills, properties

Motor function

Intelligence

Social behaviour

Body height

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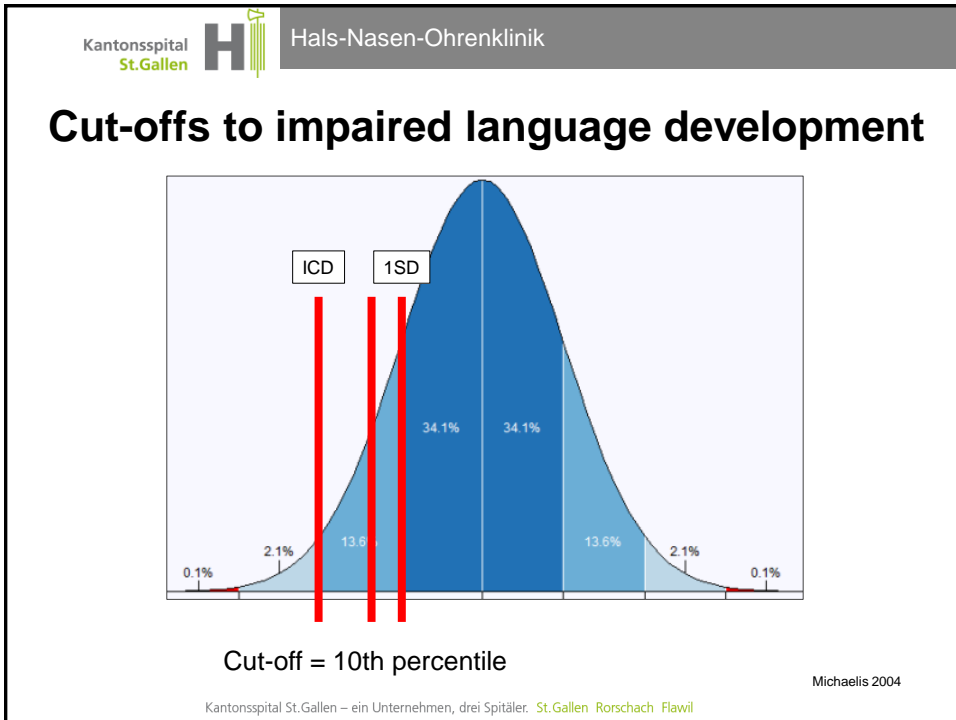
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## Normal language development

Milestone	Percentage
Below 0.1%	0.1%
0.1% - 2.1%	2.1%
2.1% - 13.6%	13.6%
13.6% - 34.1%	34.1%
34.1% - 50th percentile	34.1%
50th percentile - 34.1%	34.1%
34.1% - 13.6%	13.6%
13.6% - 2.1%	2.1%
2.1% - 0.1%	2.1%
Above 0.1%	0.1%

Milestone = 50th percentile

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### Language development (month of age)

Developmental step	Milestone (50th p.)	Cut-off (10th p.)
Syllabic doubling	8.-10.	11.-15.
Targeted mama, papa	10.-15.	18.-20.
Production > 50 words	18.	24.
Verb in second position	30.-36.	?

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## Parental questionnaires

Age (years)	0.	1.	2.	3.	...
-------------	----	----	----	----	-----

Little Ears

SBE-2-KT

SBE-3-KT

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## Specific language impairment (definition)

### Exclusion criteria:

- Hearing impairment
- Mental retardation
- Autism
- Syndrome
- Clef lip and palate
- (Bilingualism)
- et al.

### Inclusion criteria:

- Significant deficits of language skills

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## Specific language impairment (epidemiology)

Frequency 5-8 %

Gender ratio (♂ : ♀) 2-3 : 1

Tomblin et al., 1997

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## Specific language impairment (symptoms)

- Delayed first word (~24 Mo.)
- Abnormalities grammar, syntax  
(„Papa Hause kommen“)
- Auditory short term memory

## Aetiology

### Genetics vs environment




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## Genetics vs environment?

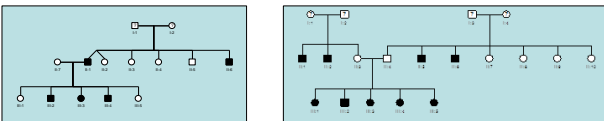
Methodology
Family aggregation studies
Twin studies
Pedigree analysis
Molecular genetics

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## Family aggregation studies

- Family aggregation of SLI
- Positive family history
  - SLI group 46 %
  - Controll group 18 %



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## Principle Twin studies

- Monozygotic twins
- 100% identical Gens
- Dizygotic twins
- ~ 50% identical gens



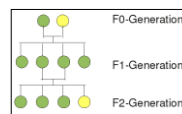
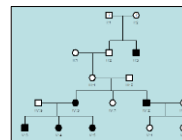
Concordance of a trait of a twin pair?

Monozygotic twins ~100 % ; dizygotic twins ~ 50% → genetics  
Monozygotic twins e.g. 30% ; dizygotic twins 30% → Environment

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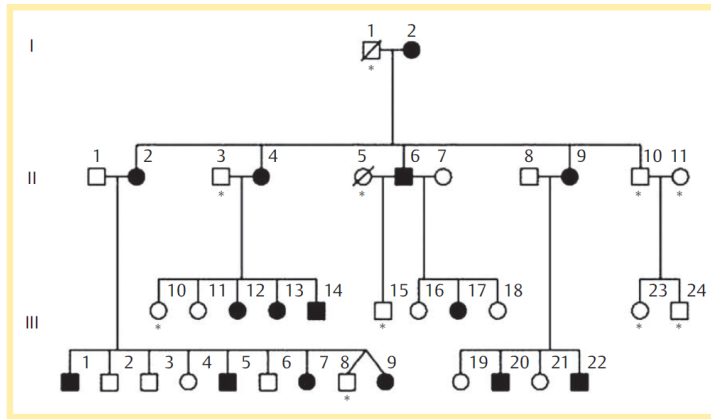
## Pedigree analysis

- Oldest method to proof heritability
- Question of mendelian inheritance
  - autosomal recessive
  - autosomal dominant
- Diseases of mendelian inheritance
  - mostly seldom
  - one gene affected



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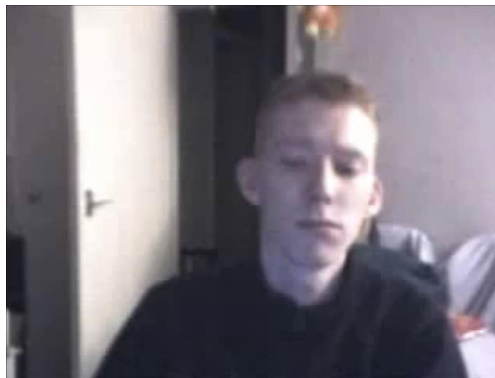
## Pedigree analysis



„KE family“, autosomal dominant inheritance Gopnik et al.,1990

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## KE family (phenotype)



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## KE family: Molekulargenetik

- Linkage analysis: gene locus 7q31 (1998)
- FOXP2*: First gene of speech and language disorder (2001)
- Specific language impairment: no alteration of *FOXP2* (2002)

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## FOXP2 and other species

- FOXP2* (715 AS): comparison human, chimpanzee, mouse
- FOXP2* in Neanderthals
- FOXP2* in knock-in mouse
  - Pitch of peeping higher
  - „braver“

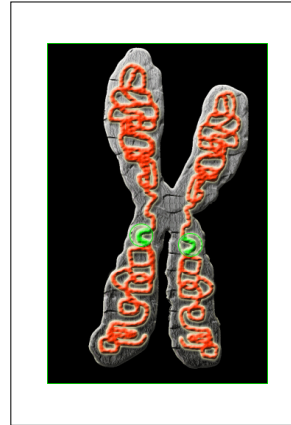
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## Molecular genetics

Linkage analysis of many SLI-families

Identification of 4 gene locus for SLI:  
16q, 19q, 13q21, 7q35-q36

1 known gene: *CNTNAP2* with  
association to SLI



## Conclusion: causes of SLI

- Multifactorial conditioned disorder
- Predominantly genetic conditioned
- Less environmental
- Interaction of several genes



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- Syndrome
- Clef lip and palate
- (Bilingualism)
- et al.

### Inclusion criteria:

- Significant deficits of language skills

## Diagnosis SLI (exclusion criteria)

- Medical history
  - Pregnancy
  - Birth
  - Bilingualism
- ENT-findings
  - Organs of articulation
  - Ears

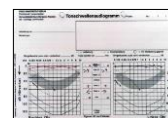
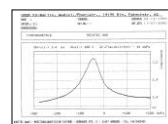
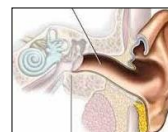


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## Diagnosis SLI (exclusion criteria)

### Exclusion of hearing impairment

- Microscopy of ears
- Flexibility of eardrum (tympanogram)
- Audiometry (pure tone audiogramm, OAE)



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## Diagnosis SLI (exclusion criteria)

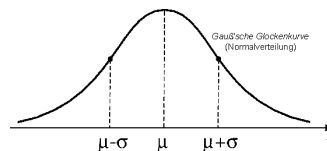
Assessment of non-verbal intelligence (CPM)

## Diagnosis SLI (inclusion criteria)

1. Informal approach (Spontaneous speech analysis)  
Subjective assessment of an examiner



2. Standardised, normative tests of language  
Validated tests with information of normal distribution





## Diagnosis SLI (Spontaneous speech analysis)

## Diagnosis SLI (comprehension)

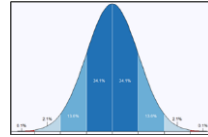


## Diagnosis SLI (grammar: singular-plural)

## Diagnosis SLI (repetition of nonsense words)

# Take home messages

- Language development is robust, wide spectrum, cut-offs variable
- Assessment of language development in children is simple
- Specific language impairment genetically conditioned
- Causes of language disorders need to be clarified to receive the correct treatment!



# Thank you!

