



## Spastische Spinalparalysen Bericht aus Tübingen

Rebecca Schüle

Hertie-Institute for Clinical Brain Research  
Junior Research Group Spastic Paraplegia  
Tübingen, Germany



## Die Kliniker



Ludger Schöls



Rebecca Schüle



Kathrin Karle



Sarah Wiethoff



Marion Döbler-  
Neumann





1

Klinik

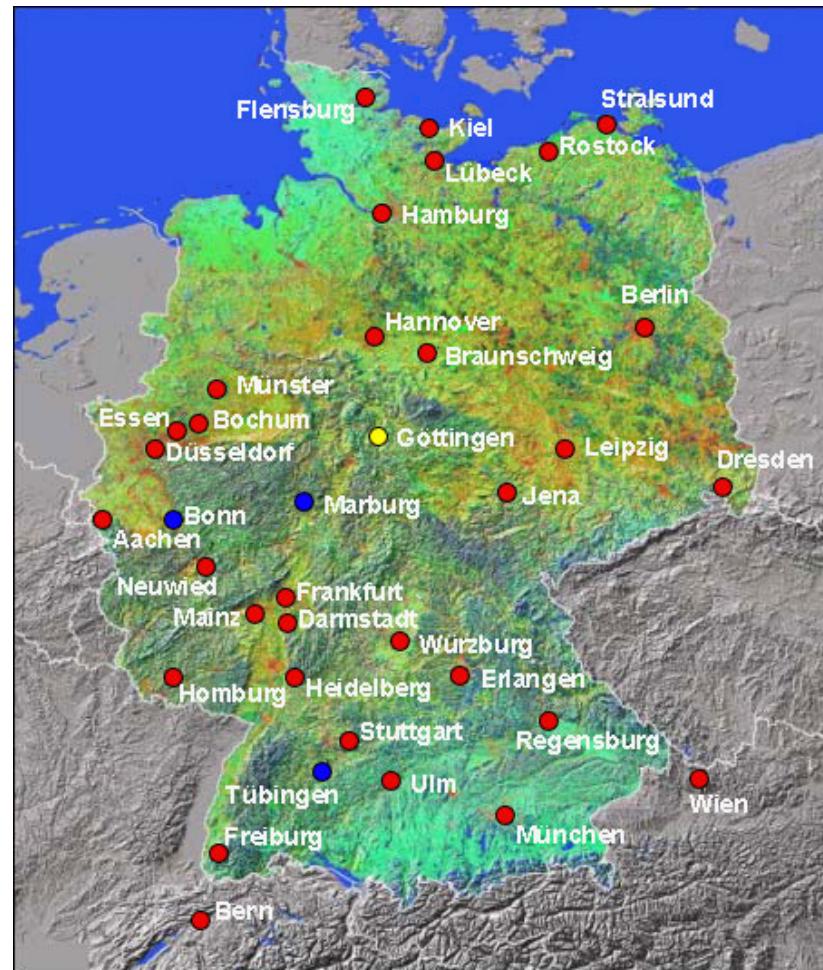


# GeNe *Move*



German Network of  
Hereditary Movement Disorders

Deutsches Netzwerk für  
erbliche Bewegungsstörungen





- „Was erwartet mich?“
- „Sitze ich irgendwann im Rollstuhl?“
- „Was kommt außer dem Gehen noch dazu?“
- „Bei mir ist die Krankheit erst spät ausgebrochen. Wenn ich Kinder habe, wird das dann auch so sein?“
- „Ich habe Erektionsstörungen. Gehört das auch zur HSP?“
- „Mein Kind hat eine SPG11. Müssen wir damit rechnen, dass auch die kognitiven Fähigkeiten schlechter werden? Wird es einen Schulabschluss machen können?“
- ...





Patient name \_\_\_\_\_  
Rater \_\_\_\_\_ Date \_\_\_\_\_

H S P-  
\_\_\_\_\_  
Including center - Family number - Individual number

**1) Walking distance without pause**

*Due to history, walking aids allowed*

- 0: Normal, unlimited
- 1: Abnormal exhaustion due to spasticity after more than 500m
- 2: Walking distance less than 500m
- 3: Walking distance less than 10 m
- 4: Unable to walk

**2) Gait quality**

*Patient is asked to walk as fast as possible a 10 meter distance including one turn*

- 0: Normal
- 1: Mild stiffness, running still possible
- 2: Clearly spastic gait, interfering with running
- 3: Spastic gait requiring use of canes/walker
- 4: Unable to walk for a 10 metre distance even with maximal support

**3) Maximum gait speed** \_\_\_\_\_ sec

*Time for a 10 meter distance including one turn, taken by stop watch*

- 0: Normal
- 1: Slightly reduced (10m:  $\geq 5s$ )
- 2: Moderately reduced (10m:  $\geq 10s$ )
- 3: Severely reduced (10m:  $\geq 20s$ )
- 4: Unable to walk for a 10m distance or time  $\geq 40s$

**4) Climbing stairs**

*5 steps upstairs – turn – 5 steps downstairs*

- 0: Normal: needs no support of the banister
- 1: Mild impairment: needs intermittent support of the banister
- 2: Moderate impairment: needs permanent support of the banister
- 3: Severe impairment: needs support of another person or additional walking aid to perform task
- 4: Unable to climb stairs

**5) Speed of stair climbing** \_\_\_\_\_ sec

*Time for 5 steps upstairs – turn – 5 steps downstairs, taken by stop-watch*

- 0: Normal
- 1: Slightly reduced ( $\geq 5s$  to perform task)
- 2: Moderately reduced ( $\geq 10s$  to perform task)
- 3: Severely reduced ( $\geq 20s$  to perform task)
- 4: Unable to climb stairs

**6) Arising from chair**

*Patient attempts to arise from a straight-back wood or metal chair with arms folded across chest*

- 0: Normal
- 1: Slow, or may need more than one attempt.
- 2: Pushes self up from arms of seat.
- 3: Tends to fall back and may have to try more than one time but can get up without help.
- 4: Unable to arise without help.

**7) Spasticity - hip adductor muscles**

*Score more severely affected side*

- 0: No increase in muscle tone
- 1: Slight increase in muscle tone, manifested by a catch and release
- 2: More marked increase in muscle tone through most of the range of motion
- 3: Considerable increase in muscle tone – passive movement is difficult
- 4: Limb stiff in adduction

**8) Spasticity - knee flexion**

*Score more severely affected side*

- 0: No increase in muscle tone
- 1: Slight increase in muscle tone, manifested by a catch and release
- 2: More marked increase in muscle tone through most of the range of motion
- 3: Considerable increase in muscle tone – passive movement is difficult
- 4: Limb stiff in flexion or extension

**9) Weakness - hip abduction**

- 0: No weakness
- 1: Mild weakness (4/5)
- 2: Moderate weakness (3/5)
- 3: Severe weakness (1-2/5)
- 4: Plegia (0/5)

**10) Weakness - foot dorsiflexion**

- 0: No weakness
- 1: Mild weakness (4/5)
- 2: Moderate weakness (3/5)
- 3: Severe weakness (1-2/5)
- 4: Plegia (0/5)

**11) Contractures of lower limbs**

*Score in supine position*

- Hip extension: lumbar spine and thighs touch the underlay
- Hip abduction: abduction up to an angle of  $>60^\circ$  between the legs possible
- Knee extension: thigh and calf touch the underlay
- Ankle dorsaextension:  $> 10^\circ$  possible
- Ankle pronation:  $> 10^\circ$  possible

- 0: No contractures
- 1: Mild, not fixed abnormal position of one joint (unilaterally or bilaterally)
- 2: Fixed contracture of one joint (unilaterally or bilaterally)
- 3: Fixed contracture of two joints (unilaterally or bilaterally)
- 4: Fixed contracture of more than two joints (unilaterally or bilaterally)

**12) Pain due to HSP related symptoms**

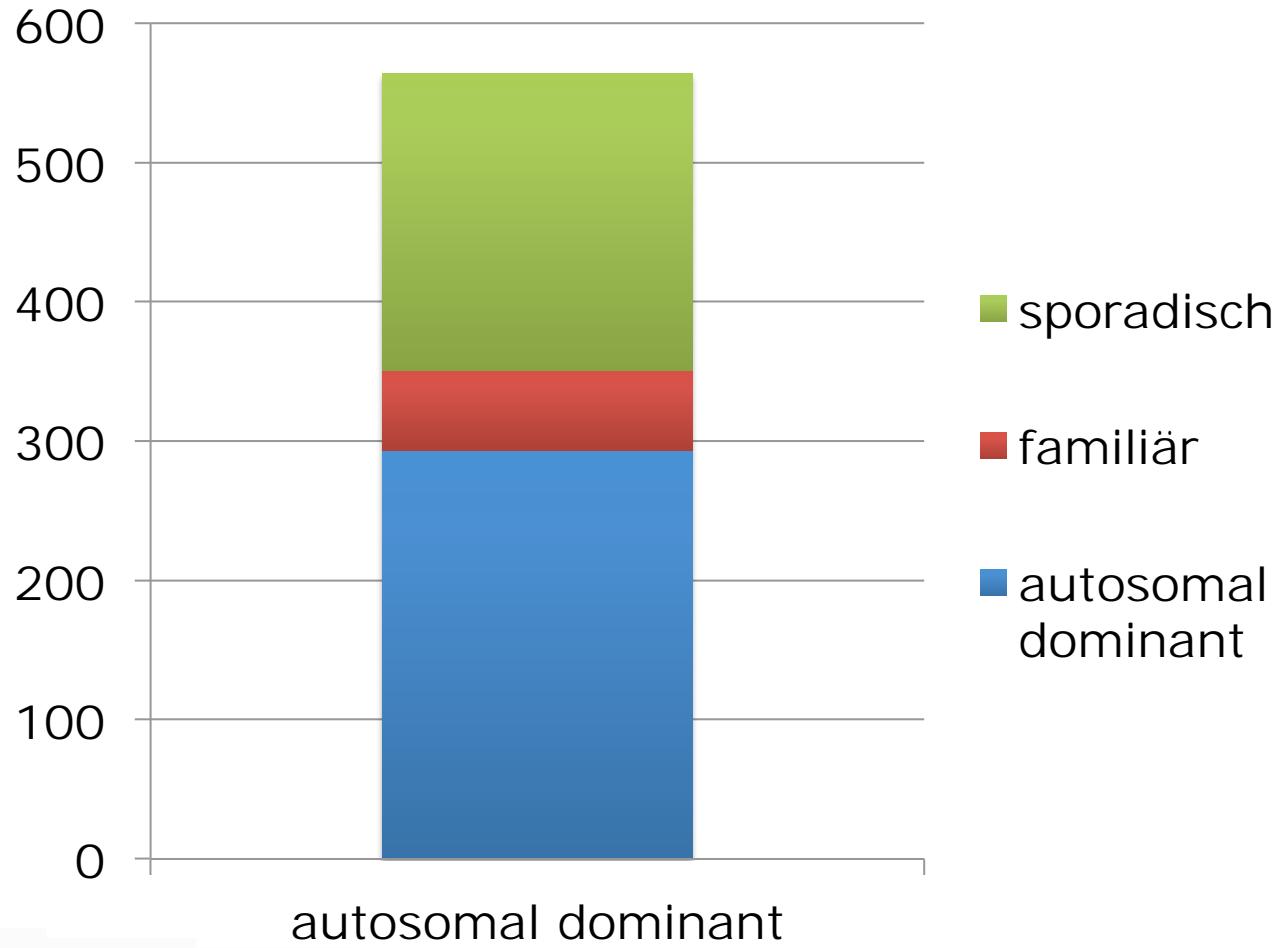
- 0: None
- 1:  $\leq 50\%$  of waking day present AND intensity 0 – 3 points on visual analogue scale
- 2:  $\leq 50\%$  of waking day present AND intensity 4 – 10 points on visual analogue scale
- 3:  $> 50\%$  of waking day present AND intensity 0 – 3 on visual analogue scale
- 4:  $> 50\%$  of waking day present AND intensity 4 – 10 points on visual analogue scale

**13) Bladder and bowel function**

- 0: Normal bladder and bowel function
- 1: Urinary or fecal urgency (difficulties to reach toilet in time)
- 2: Rare and mild urge incontinence (no nappy required)
- 3: Moderate urge incontinence (requires nappy or catheter when out of the house)
- 4: Permanent catheterization or permanent nappy

**Landmarks of Disability**

- 1: able to walk >500m without walking aid
- 2: able to walk >500m with walking aid
- 3: able to walk less than 500m with walking aid
- 4: not able to walk







## 1. Querschnittstudie

Daten aus einer einmaligen Untersuchung von Betroffenen in verschiedenen Krankheitsstadien

## 2. Längsschnittstudie

Daten aus jährlichen Verlaufsuntersuchungen



German Network of  
Hereditary Movement Disorders

Deutsches Netzwerk für  
erbliche Bewegungsstörungen



EuroSPA



2003-2006  
2007-2008

2008-2011

seit 2012



## Wie können Sie uns unterstützen?

Kommen Sie regelmäßig in eine unserer Netzwerk-Ambulanzen!

HSP-Ambulanz Tübingen

Tel. 07071 29 85165

Email [Spezialambulanz.Neurologie@med.uni-tuebingen.de](mailto:Spezialambulanz.Neurologie@med.uni-tuebingen.de)

HSP-Kinderambulanz Tübingen

Tel. 07071 29 84734



## Das DZNE-Netzwerk



Rostock  
Florian Rimmеле / Christoph Kamm  
0381 – 494 - 5276

Magdeburg  
Stefan Vielhaber  
0391– 671 5001

Bonn  
Delia Kurzwelly  
0228 – 287 14431

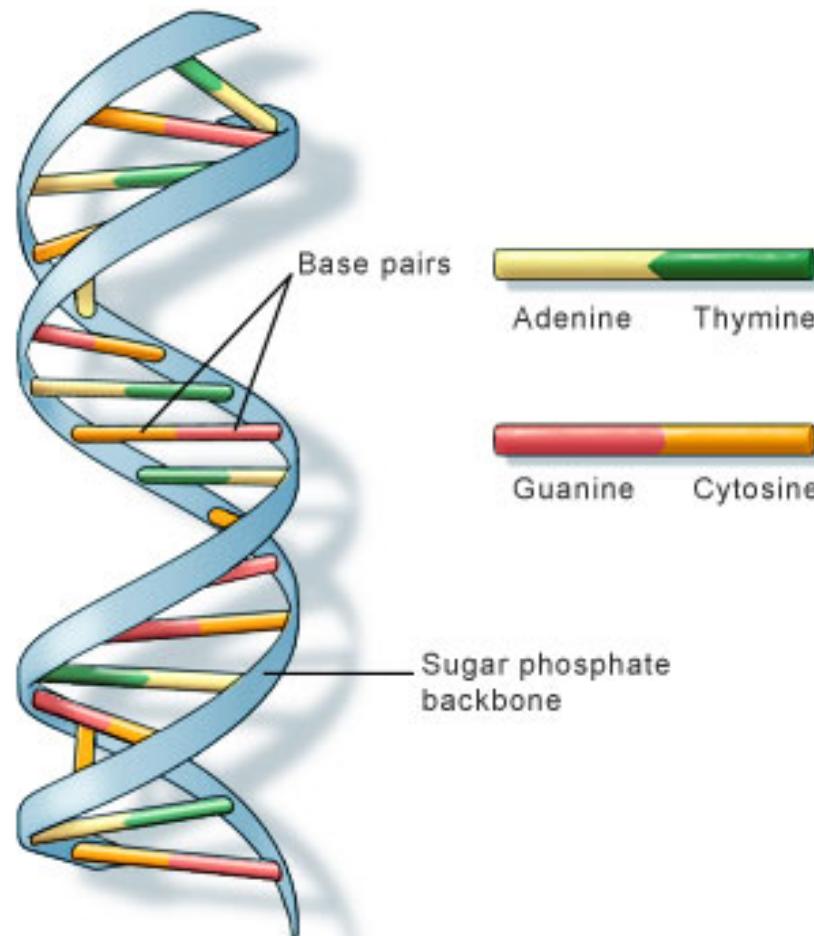
Tübingen  
Rebecca Schüle  
07071- 29 - 85165

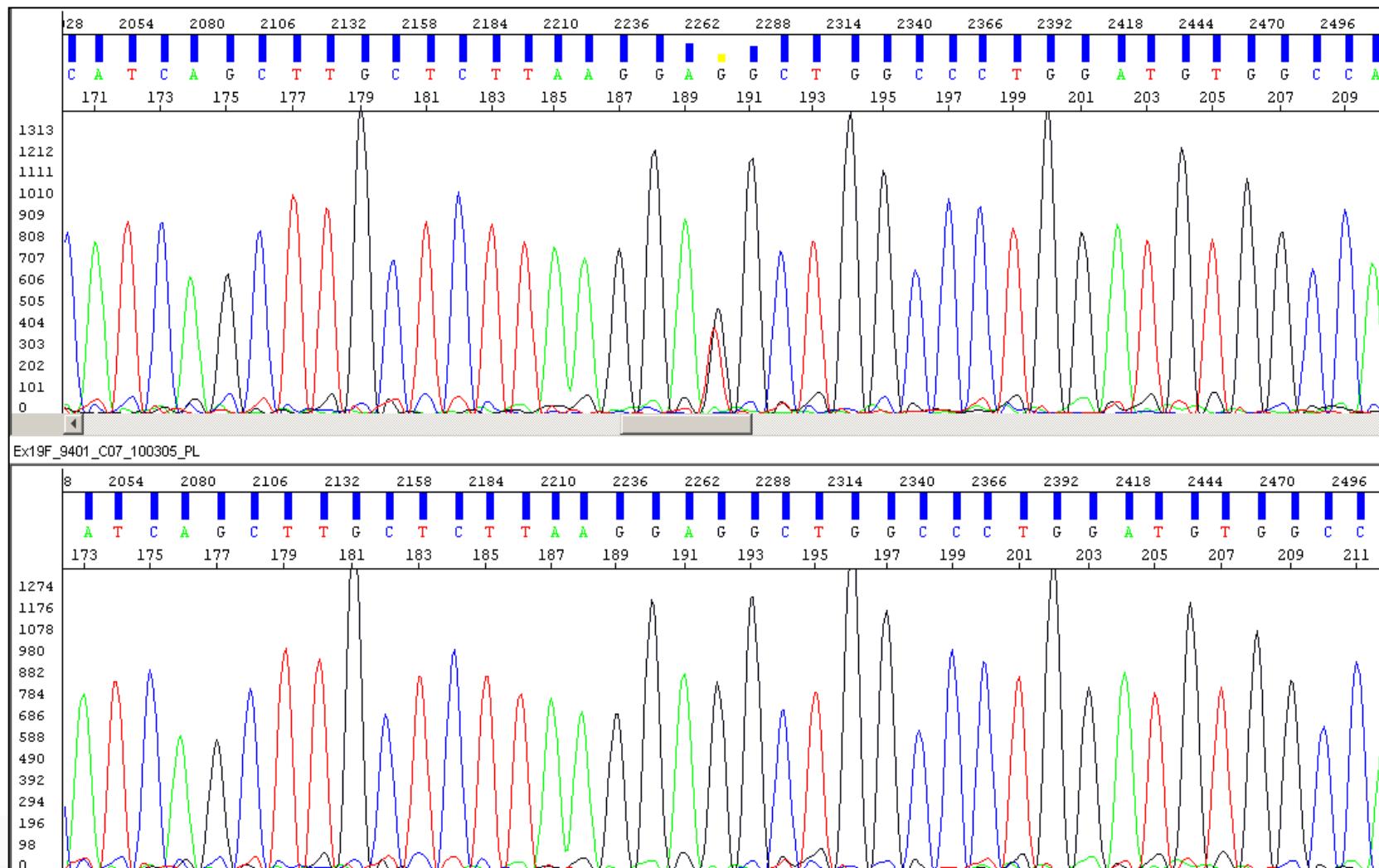
München  
Constanze Gallenmüller  
089 - 5160 - 7425



2

Genetik







■ Autosomal dominant  
■ Autosomal recessive  
■ X-linked



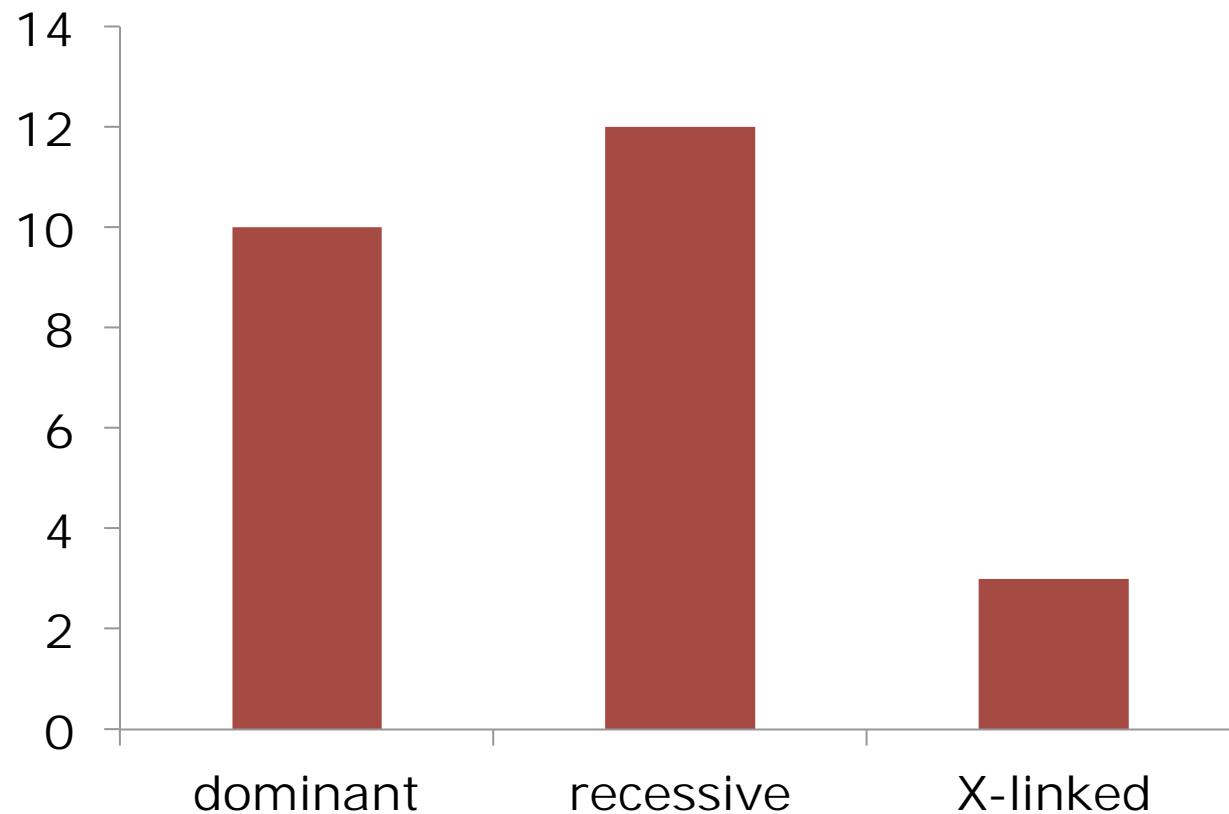
Genom ~ 3.000.000.000 Basen (3Gb)

Exom ~ 30.000.000 Basen (30Mb)

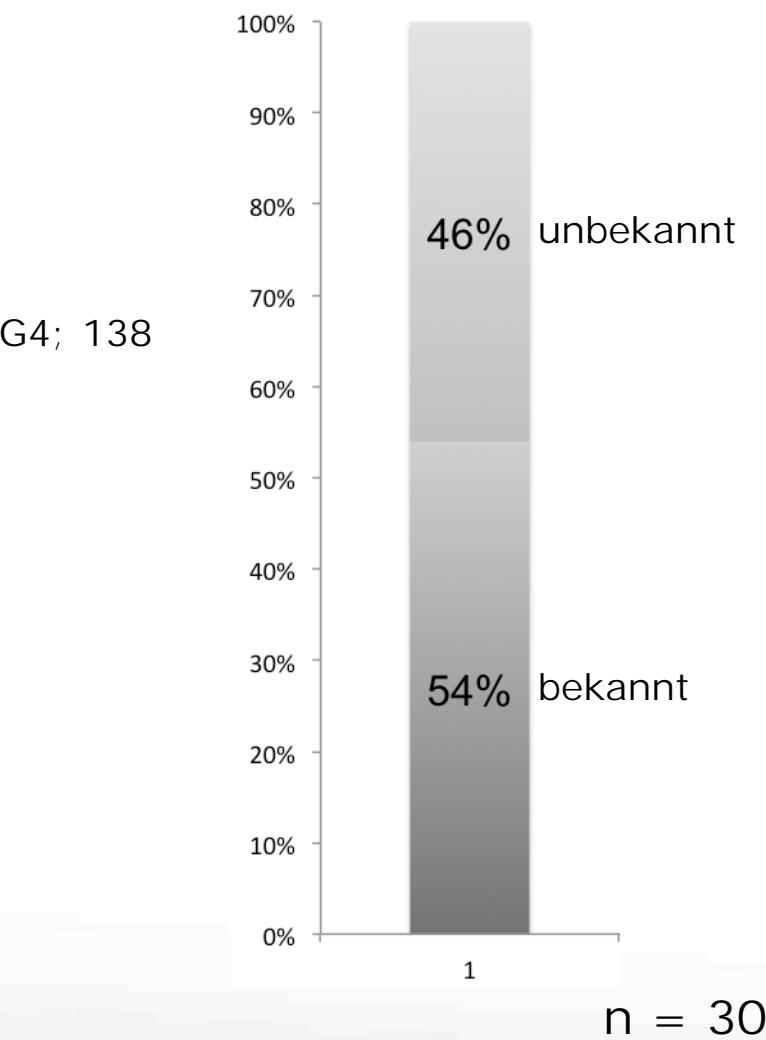
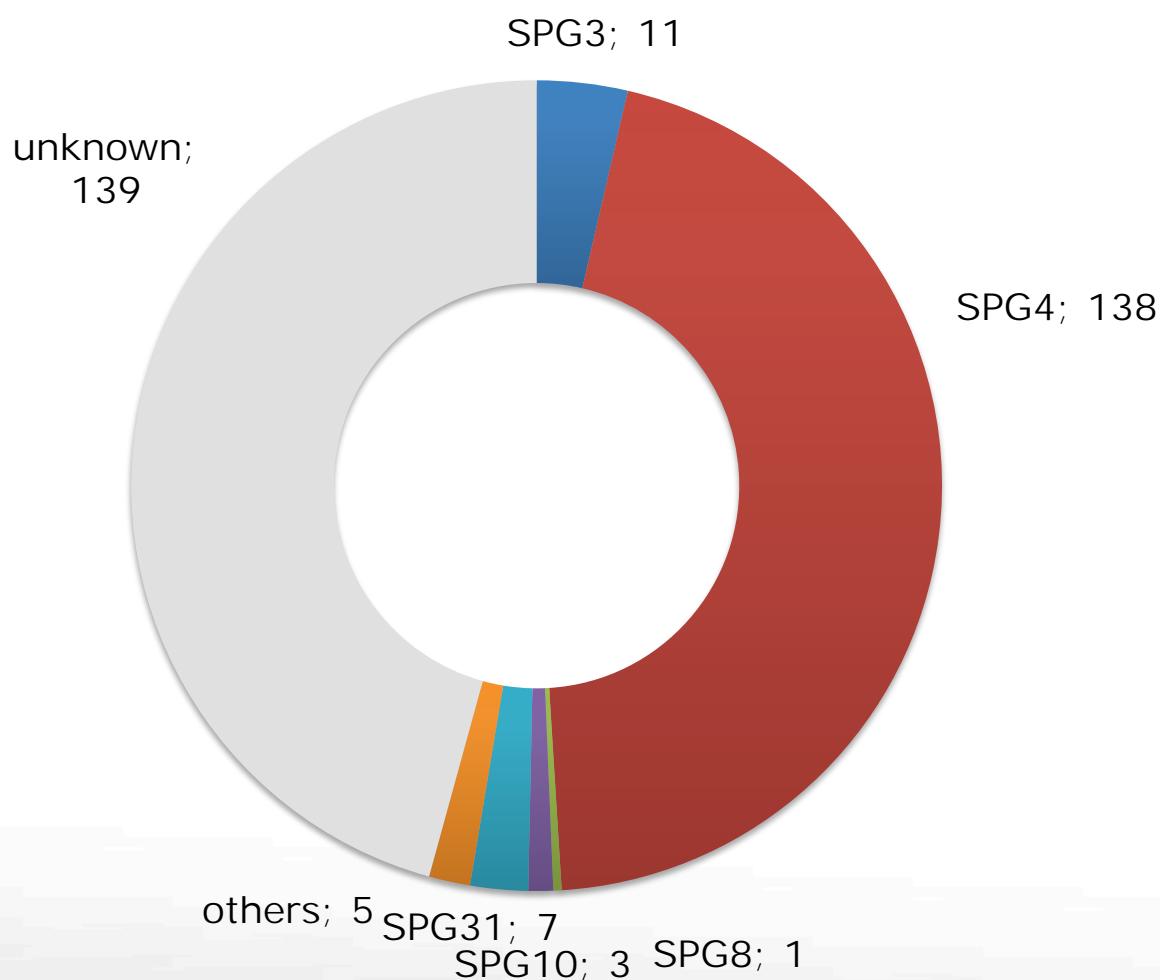
HSP-Gene ~ 30.000 Basen (30kb)



# Zu viele Gene!



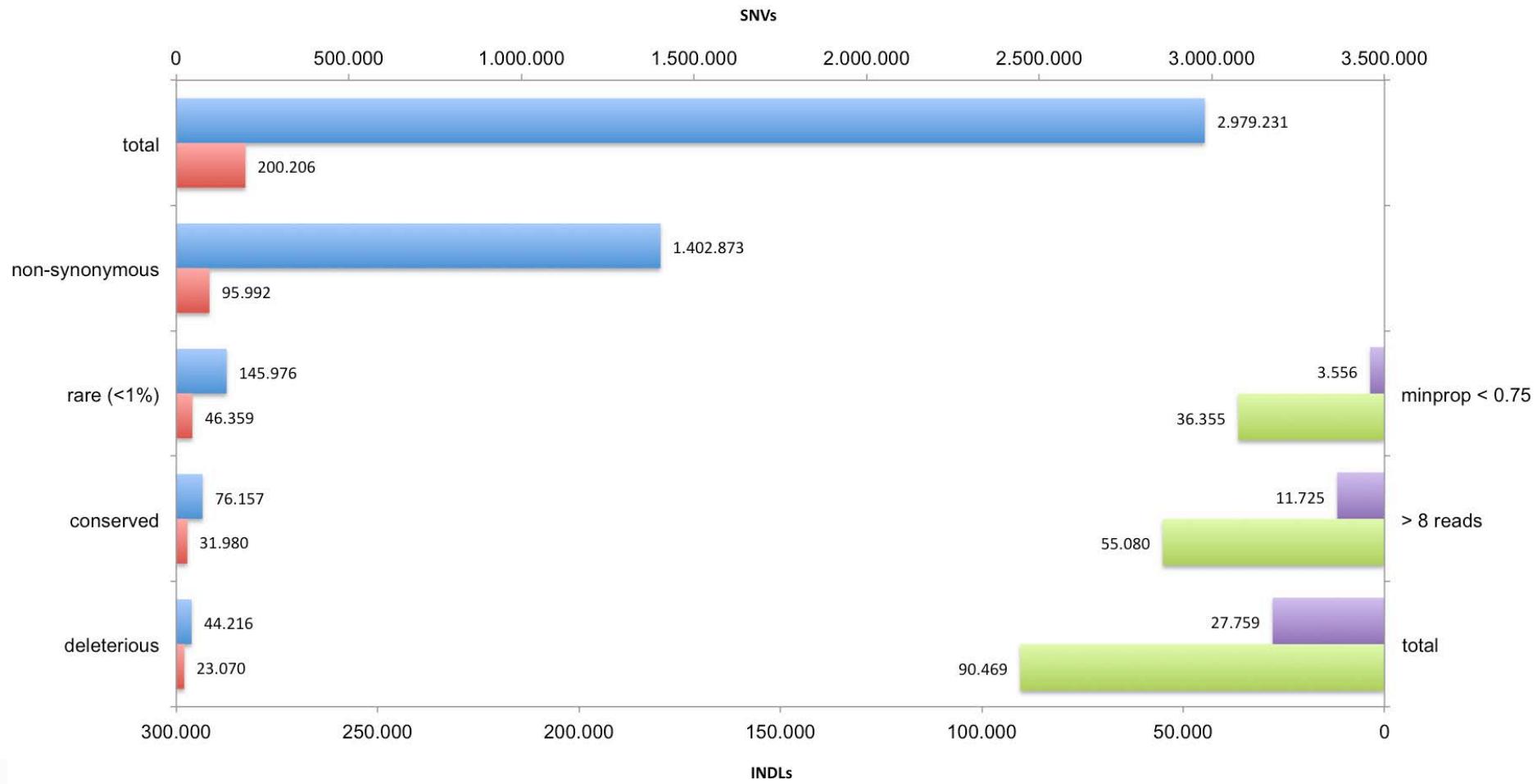
## Häufigkeit verschiedener dominanter HSP-Untertypen

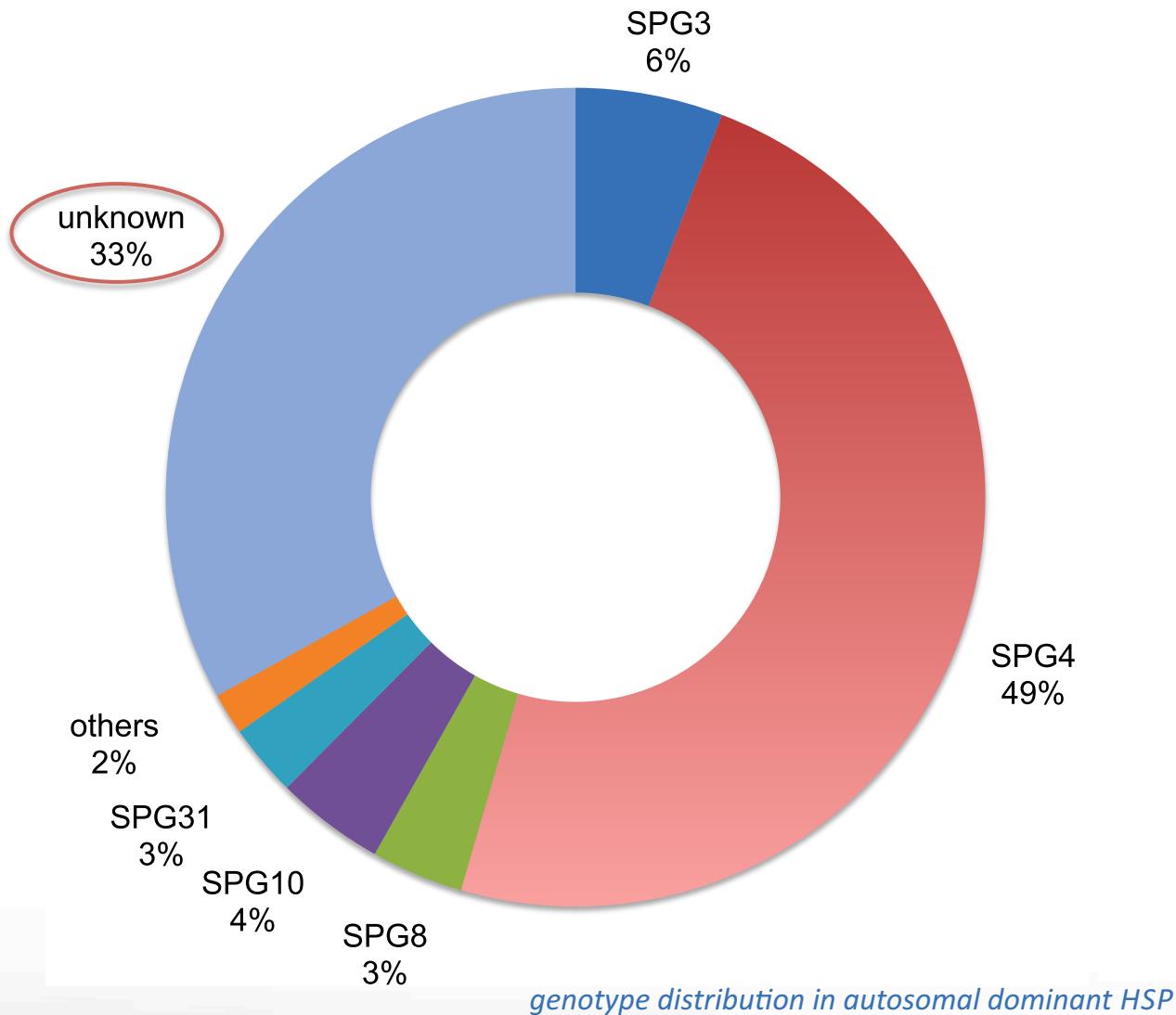






	Index	Total
Dominant	83	98
Familiär	26	30
Sporadisch	13	13
Gesamt	122	141







Wozu brauchen wir denn all  
die HSP-Gene?



# Woher kommt die Therapie?



3

Therapie



# Was ist FES?

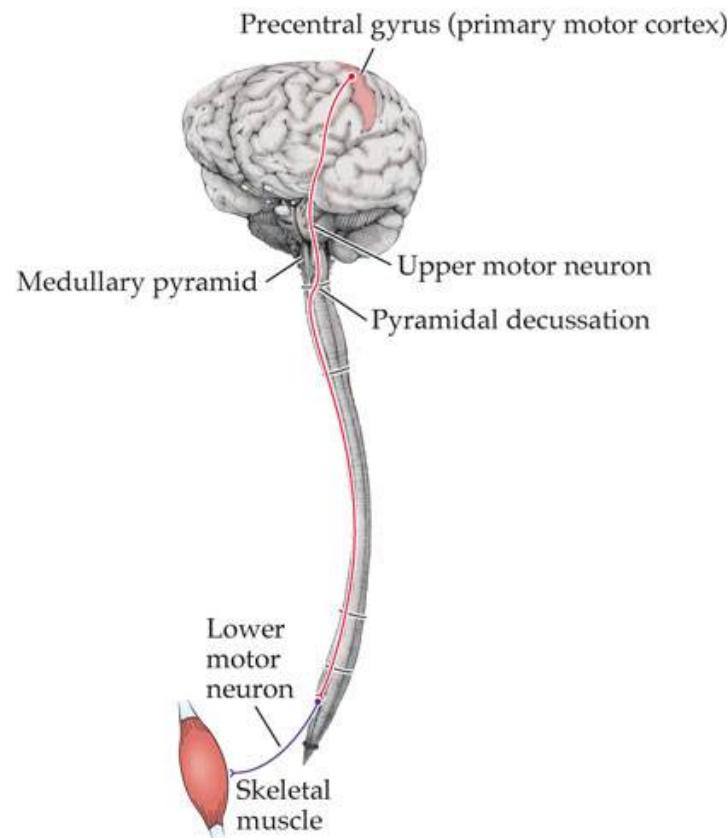
FES – funktionelle Elektrostimulation

Durch elektrische Stimulation eines Nerven wird eine Muskelkontraktion und damit eine Bewegung ausgelöst.

Bisher v.a. bei Schlaganfall und Multipler Sklerose eingesetzt.

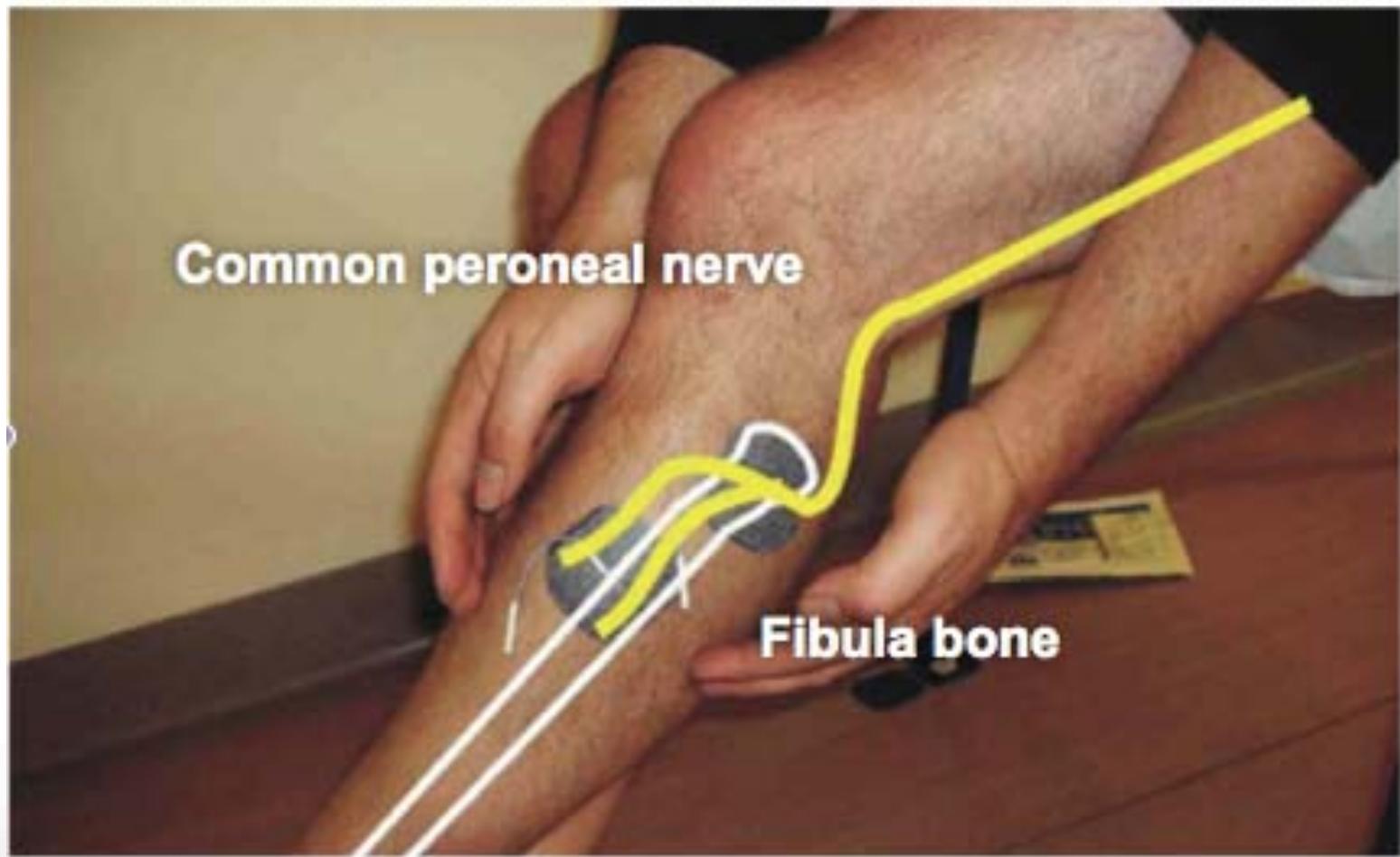


# Warum wirkt FES bei der HSP?





# Die Behandlung des Fallfußes





# Die FES-Studie in Tübingen

50 Probanden werden zufällig in eine Behandlungsgruppe und eine Kontrollgruppe eingeteilt.

Wer kann teilnehmen?

HSP, Gehstrecke >100m, kein Botox (!), kein Herzschrittmacher

Probanden in der Behandlungsgruppe erhalten für 6 Monate 2 Stimulatoren.



# Wie läuft die Studie ab?

	Visit 1	Visit 1*	Visit 2	Visit 3
	Tag 1	Tag 2	Woche 6	Woche 24
		<i>Nur FES-Gruppe</i>		
10m walk	X		X	X
PCI	X		X	X
6 min endurance	X		X	X
PIADS	X			X
Video-Ganganalyse	X			X
Sturztagebuch	Austeilen			Einsammeln
Sonstiges	Randomisierung	Prüfung der Stimulator-einstellungen	Prüfung der Stimulator-einstellungen	



## Stand der Rekrutierung

13.10.10

20.10.11

22.04.12

29.08.12

1

3

5

7

9

11

13

15

17

19

21

23

25

27

29

31

33

35

37

39

41

43

45

47

49

51

53

25

42

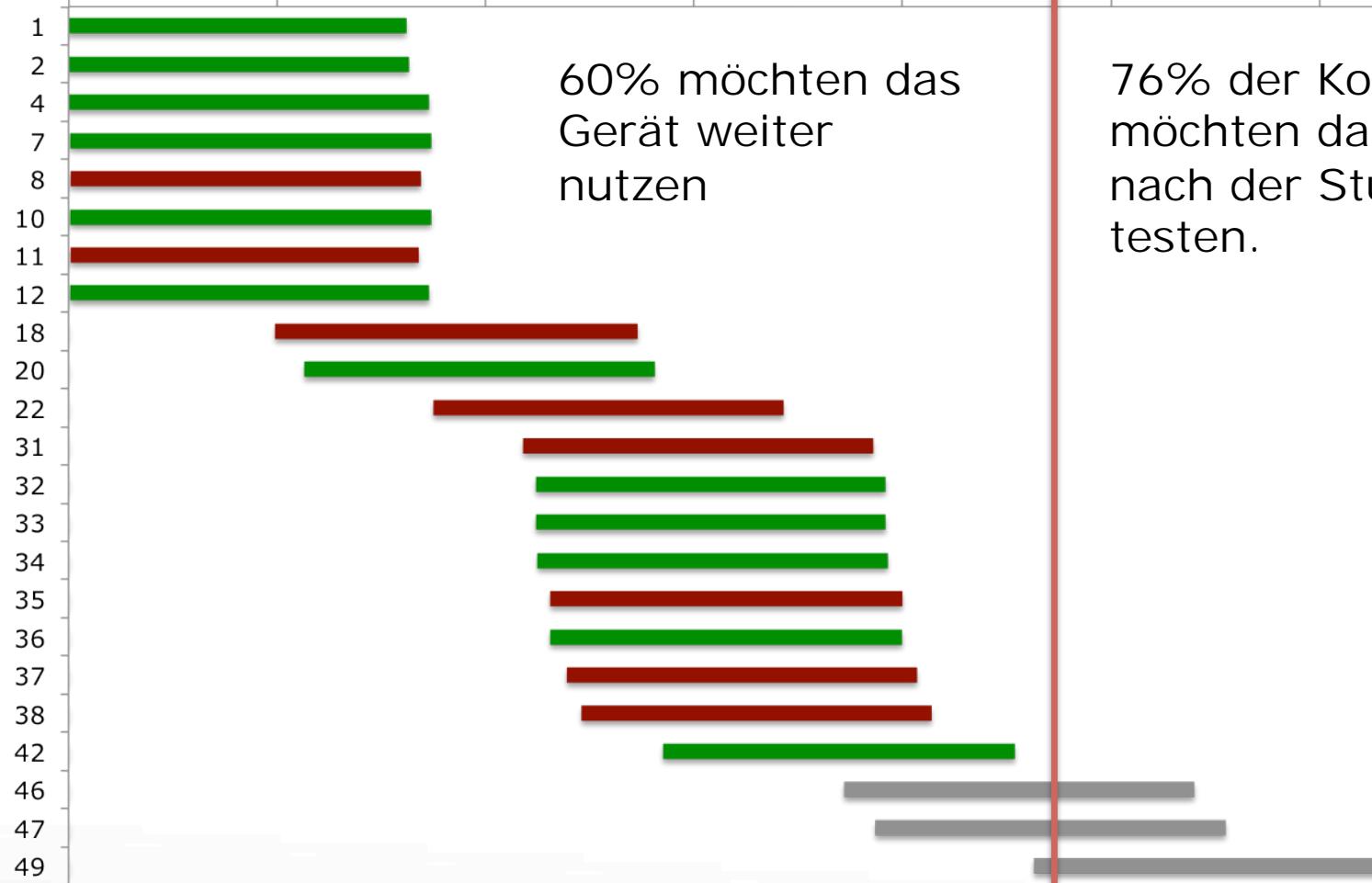


## FES-Gruppe

13.10.10

22.04.12

29.08.12





## Verbesserte Verordnungsmöglichkeiten über die Krankenkasse

### Netzwerk an FES-Ambulanzen

- Physiotherapeuten
- Neurologen
- (Sanitätshäuser)



Ludger Schöls



Rebecca Schüle



Kathrin Karle



Sarah Wiethoff



Marion Döbler-  
Neumann