

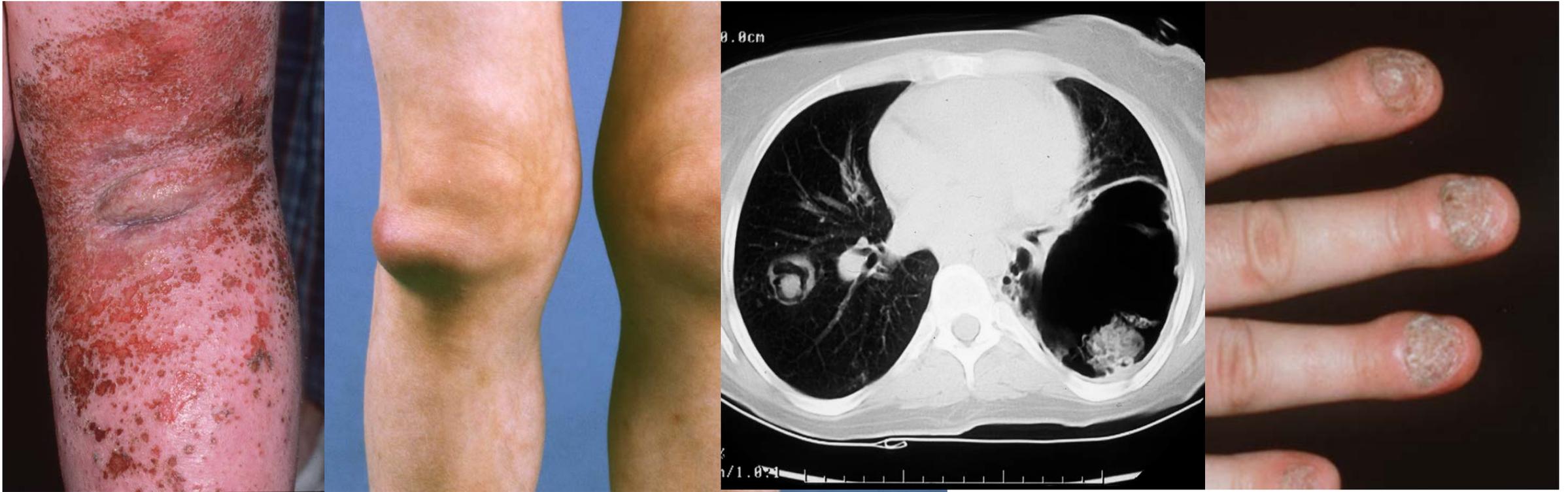
**UNIVERSITÄTS
KLINIKUM** FREIBURG

Die Lebensqualität von Patienten mit STAT3-Hyper-IgE Syndrom Bodo Grimbacher und Eyad Jannoud

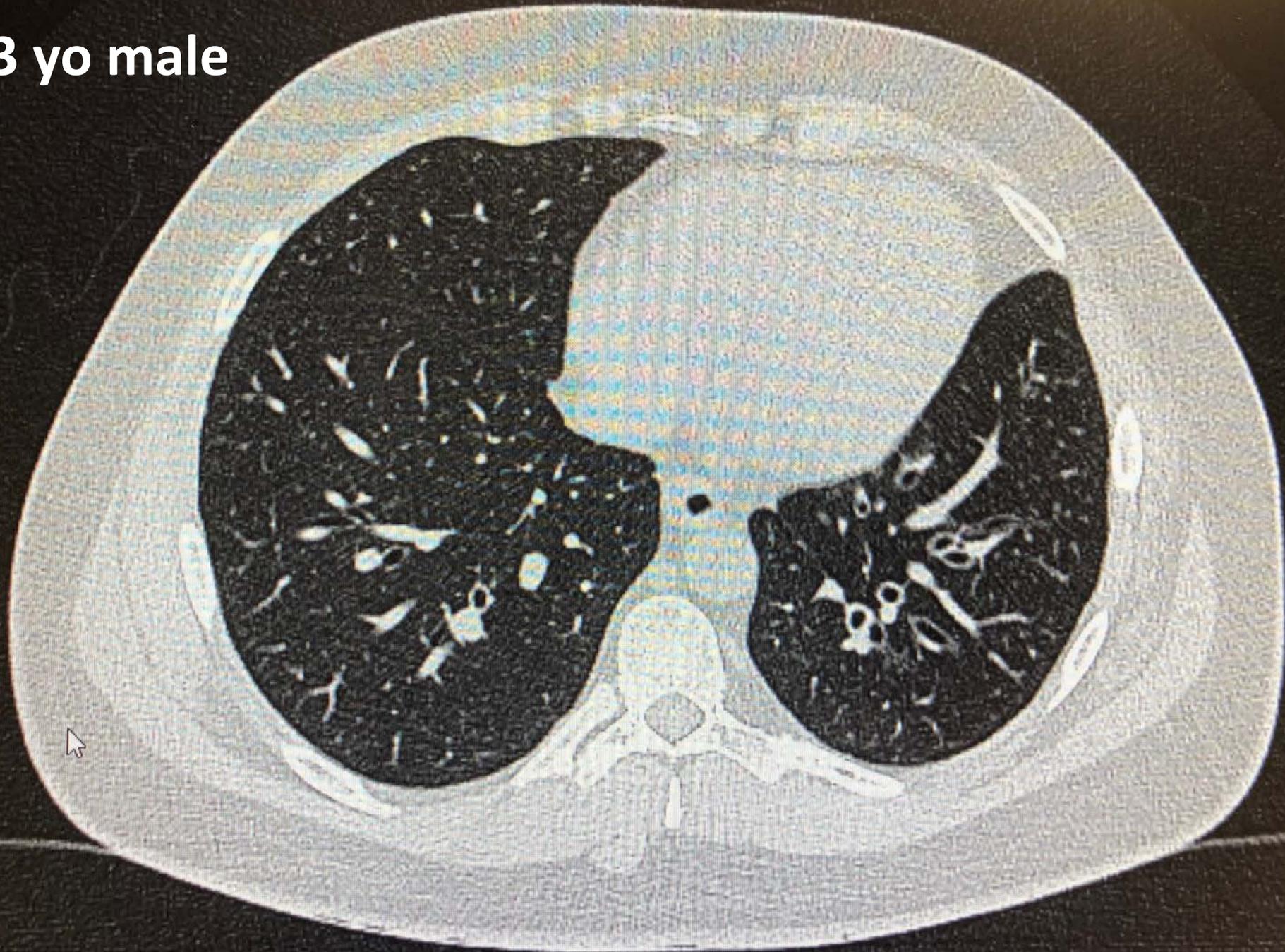
bodo.grimbacher@uniklinik-freiburg.de

eyad.jannoud@uniklinik-freiburg.de

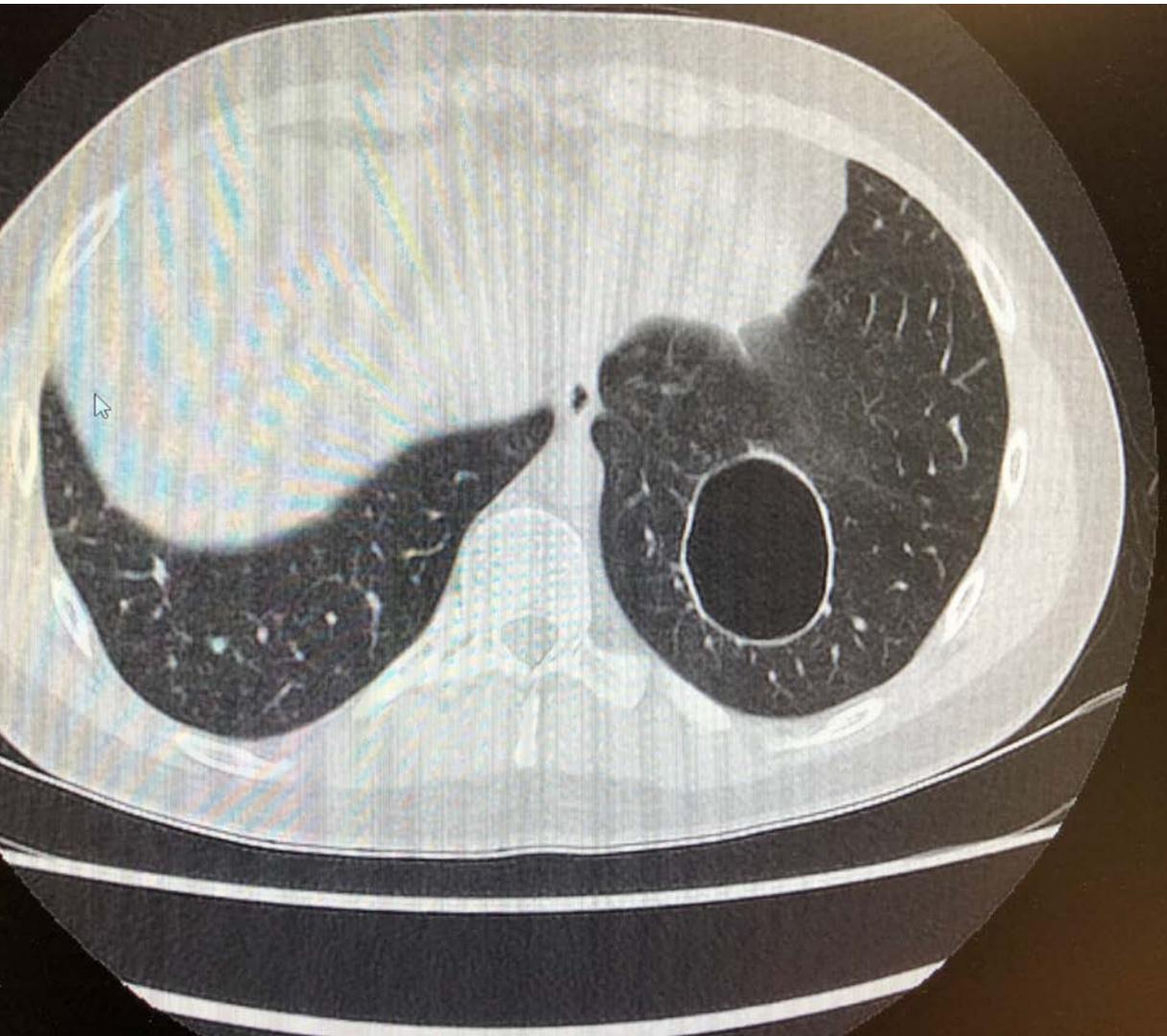
Warum sollten wir die Lebensqualität von Patienten mit DN-STAT3 Mutationen erheben?



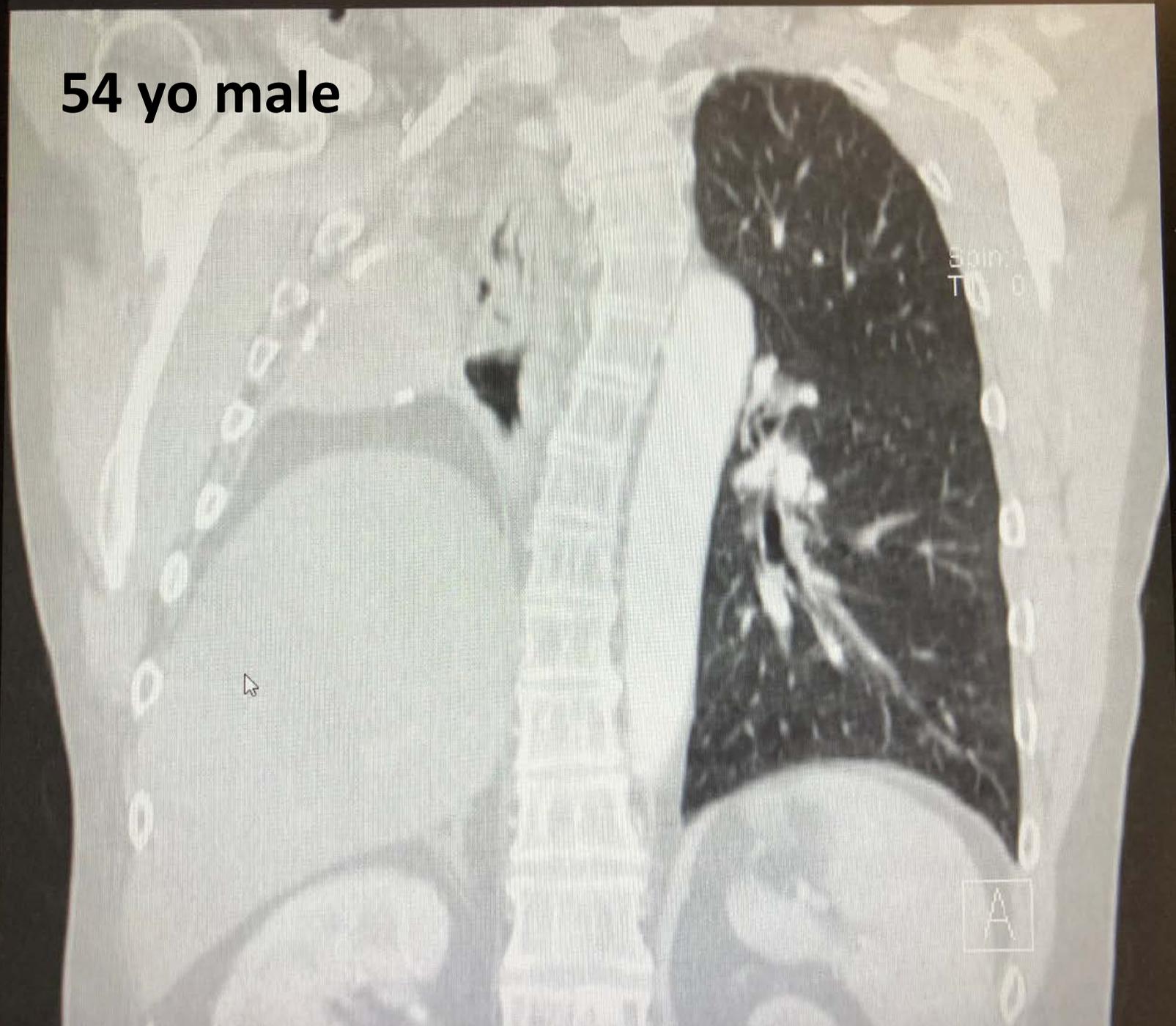
33 yo male



26 yo male



54 yo male



Methoden

- 16 erwachsene Patienten und Patientinnen mit STAT3-LOF Mutation und 22 betreuende Ärzte und Ärztinnen erhielten:
 - The FACIT Fatigue Scale
 - The Dermatology Life Quality Index (DLQI)
 - The St. Georges Respiratory Questionnaire (SGRQ)
- Bisher haben 25 Patienten*innen geantwortet

FACIT Fatigue Scale (Version 4)

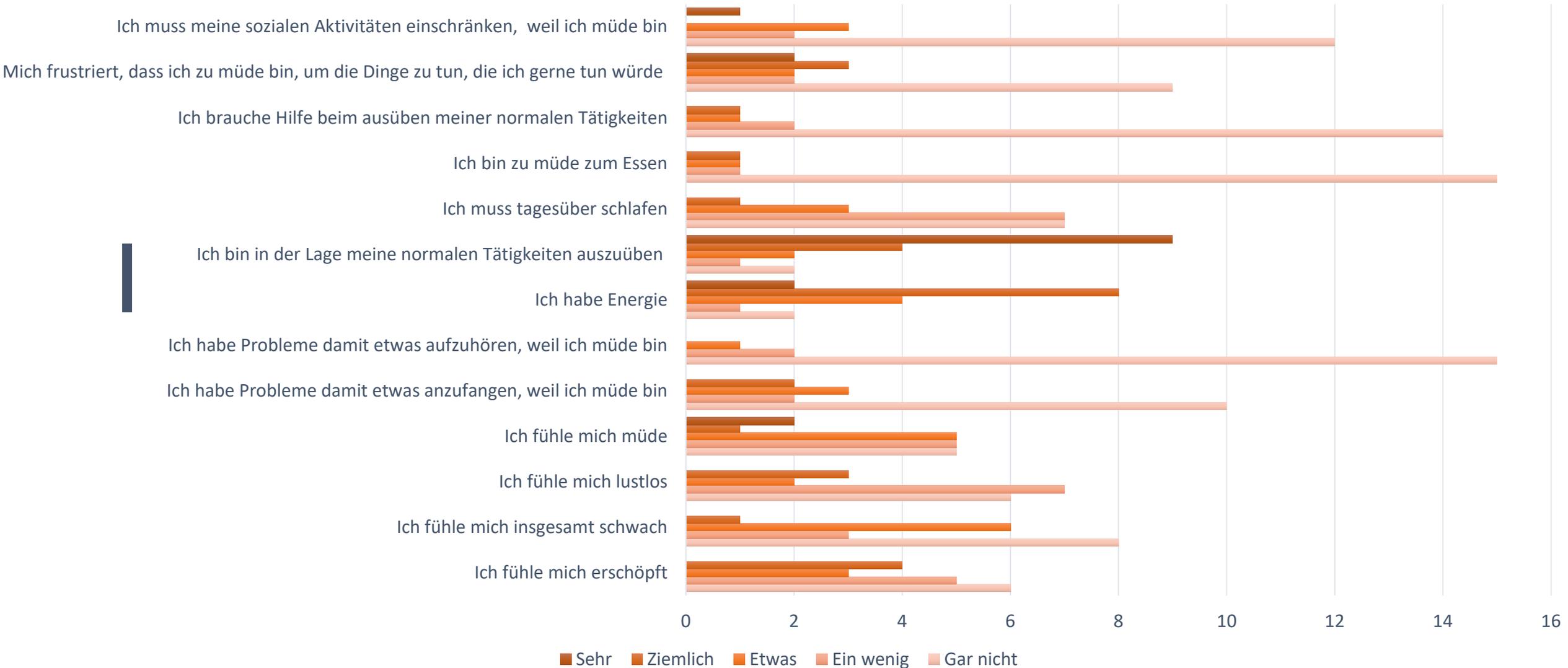
Below is a list of statements that other people with your illness have said are important. **Please circle or mark one number per line to indicate your response as it applies to the past 7 days.**

		Not at all	A little bit	Some- what	Quite a bit	Very much
HI7	I feel fatigued	0	1	2	3	4
HI12	I feel weak all over	0	1	2	3	4
An1	I feel listless (“washed out”)	0	1	2	3	4
An2	I feel tired.....	0	1	2	3	4
An3	I have trouble <u>starting</u> things because I am tired.....	0	1	2	3	4
An4	I have trouble <u>finishing</u> things because I am tired	0	1	2	3	4
An5	I have energy.....	0	1	2	3	4
An7	I am able to do my usual activities.....	0	1	2	3	4
An8	I need to sleep during the day	0	1	2	3	4
An12	I am too tired to eat.....	0	1	2	3	4
An14	I need help doing my usual activities	0	1	2	3	4
An15	I am frustrated by being too tired to do the things I want to do.....	0	1	2	3	4
An16	I have to limit my social activity because I am tired.....	0	1	2	3	4

Fatigue Scale

Bezogen auf die letzten 7 Tage

(n=18)



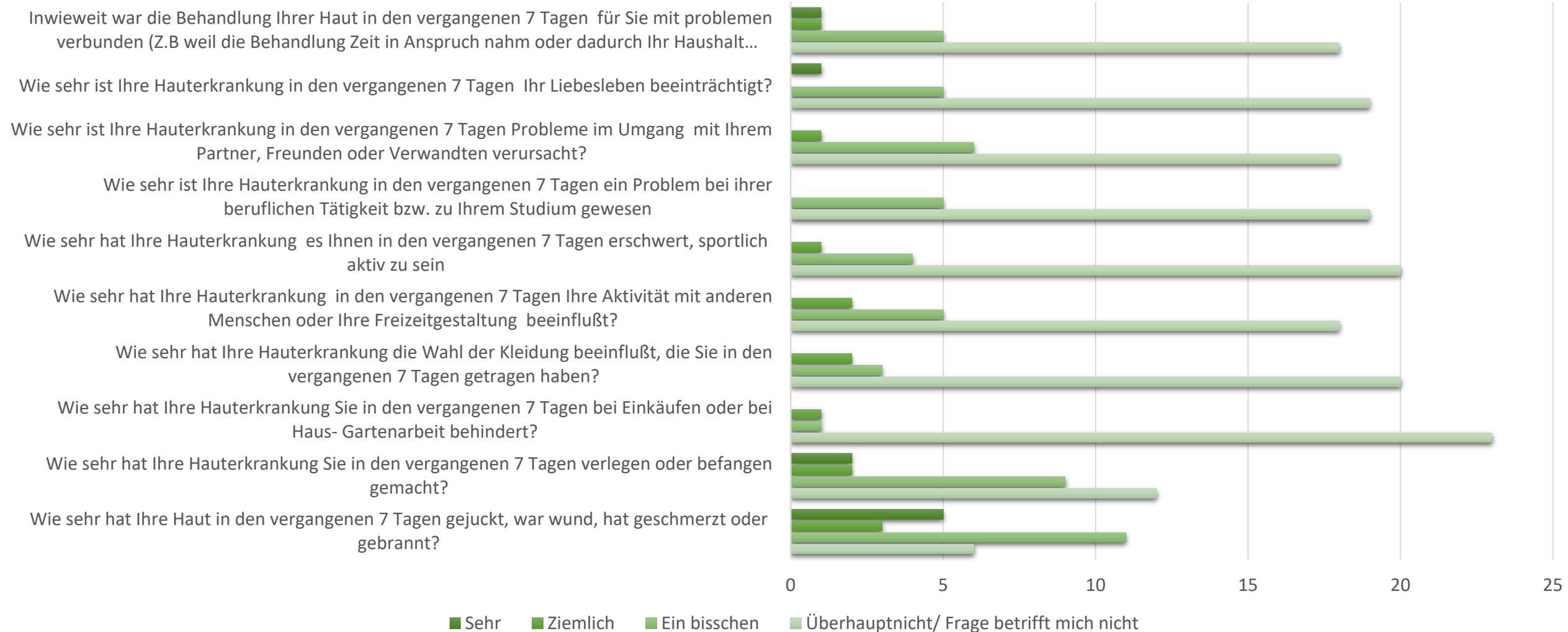
DERMATOLOGY LIFE QUALITY INDEX (DLQI)

The aim of this questionnaire is to measure how much your skin problem has affected your life
OVER THE LAST WEEK. Please tick (✓) one box for each question.

- | | | | |
|--|-------------------------------------|---------------------------------------|--|
| 1. Over the last week, how itchy, sore, painful or stinging has your skin been? | Very much <input type="checkbox"/> | | |
| | A lot <input type="checkbox"/> | | |
| | A little <input type="checkbox"/> | | |
| | Not at all <input type="checkbox"/> | | |
| 2. Over the last week, how embarrassed or self conscious have you been because of your skin? | Very much <input type="checkbox"/> | | |
| | A lot <input type="checkbox"/> | | |
| | A little <input type="checkbox"/> | | |
| | Not at all <input type="checkbox"/> | | |
| 3. Over the last week, how much has your skin interfered with you going shopping or looking after your home or garden? | Very much <input type="checkbox"/> | | |
| | A lot <input type="checkbox"/> | | |
| | A little <input type="checkbox"/> | | |
| | Not at all <input type="checkbox"/> | Not relevant <input type="checkbox"/> | |
| 4. Over the last week, how much has your skin influenced the clothes you wear? | Very much <input type="checkbox"/> | | |
| | A lot <input type="checkbox"/> | | |
| | A little <input type="checkbox"/> | | |
| | Not at all <input type="checkbox"/> | Not relevant <input type="checkbox"/> | |
| 5. Over the last week, how much has your skin affected any social or leisure activities? | Very much <input type="checkbox"/> | | |
| | A lot <input type="checkbox"/> | | |
| | A little <input type="checkbox"/> | | |
| | Not at all <input type="checkbox"/> | Not relevant <input type="checkbox"/> | |
| 6. Over the last week, how much has your skin made it difficult for you to do any sport? | Very much <input type="checkbox"/> | | |
| | A lot <input type="checkbox"/> | | |
| | A little <input type="checkbox"/> | | |
| | Not at all <input type="checkbox"/> | Not relevant <input type="checkbox"/> | |
| 7. Over the last week, has your skin prevented you from working or studying? | Yes <input type="checkbox"/> | | |
| | No <input type="checkbox"/> | Not relevant <input type="checkbox"/> | |
| If "No", over the last week how much has your skin been a problem at work or studying? | A lot <input type="checkbox"/> | | |
| | A little <input type="checkbox"/> | | |
| | Not at all <input type="checkbox"/> | | |
| 8. Over the last week, how much has your skin created problems with your partner or any of your close friends or relatives? | Very much <input type="checkbox"/> | | |
| | A lot <input type="checkbox"/> | | |
| | A little <input type="checkbox"/> | | |
| | Not at all <input type="checkbox"/> | Not relevant <input type="checkbox"/> | |
| 9. Over the last week, how much has your skin caused any sexual difficulties? | Very much <input type="checkbox"/> | | |
| | A lot <input type="checkbox"/> | | |
| | A little <input type="checkbox"/> | | |
| | Not at all <input type="checkbox"/> | Not relevant <input type="checkbox"/> | |
| 10. Over the last week, how much of a problem has the treatment for your skin been, for example by making your home messy, or by taking up time? | Very much <input type="checkbox"/> | | |
| | A lot <input type="checkbox"/> | | |
| | A little <input type="checkbox"/> | | |
| | Not at all <input type="checkbox"/> | Not relevant <input type="checkbox"/> | |

Dermatologische Lebensqualität

Einfluss der Hauterkrankung auf die Lebensqualität von HIES-Patienten in den letzten 7 Tagen (n=25)



St. George's Respiratory Questionnaire

Questions about how much chest trouble you have had over the past 3 months.

Please tick (✓) one box for each question:

- | | most
days
a week | several
days
a week | a few
days
a month | only with
chest
infections | not
at
all |
|---|--------------------------|---------------------------|--------------------------|----------------------------------|--------------------------|
| 1. Over the past 3 months, I have coughed: | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |
| 2. Over the past 3 months, I have brought up phlegm (sputum): | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |
| 3. Over the past 3 months, I have had shortness of breath: | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |
| 4. Over the past 3 months, I have had attacks of wheezing: | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |
| 5. During the past 3 months how many severe or very unpleasant attacks of chest trouble have you had? | | | | | |

Please tick (✓) one:

- more than 3 attacks
3 attacks
2 attacks
1 attack
no attacks

6. How long did the worst attack of chest trouble last?
(Go to question 7 if you had no severe attacks)

Please tick (✓) one:

- a week or more
3 or more days
1 or 2 days
less than a day

7. Over the past 3 months, in an average week, how many good days (with little chest trouble) have you had?

Please tick (✓) one:

- No good days
1 or 2 good days
3 or 4 good days
nearly every day is good
every day is good

8. If you have a wheeze, is it worse in the morning?

Please tick (✓) one:

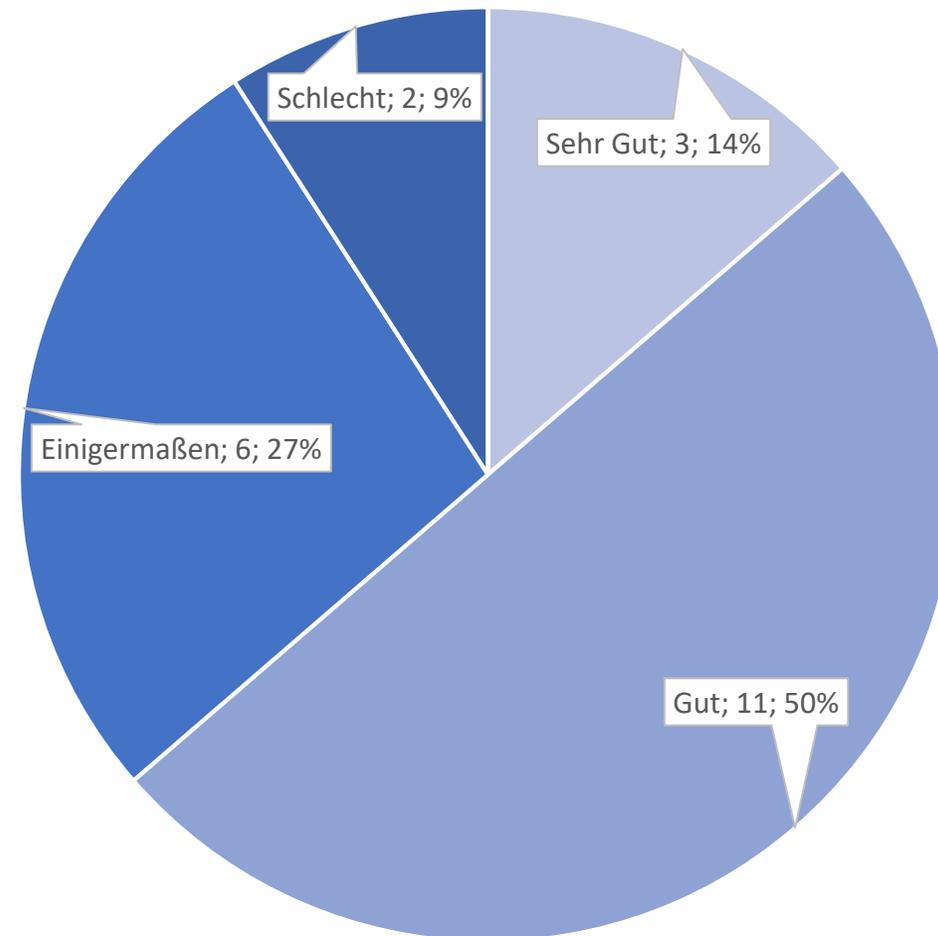
- No
Yes

Wie würden Sie Ihren derzeitigen Gesundheitszustand bewerten?

Bevor Sie den restlichen Fragebogen ausfüllen:

Bitte kreuzen Sie die Beschreibung an, die nach Ihrer Beurteilung Ihrem jetzigen Gesundheitszustand entspricht:

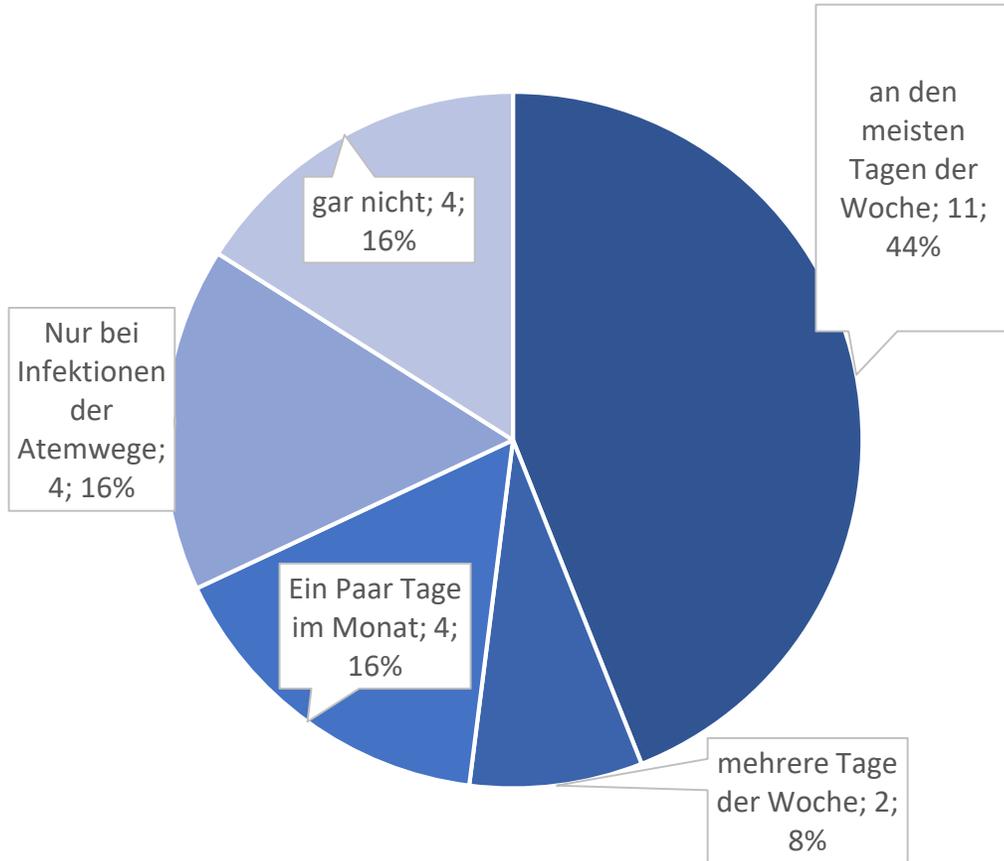
Sehr gut Gut Einigermaßen Schlecht Sehr schlecht



(n=25)

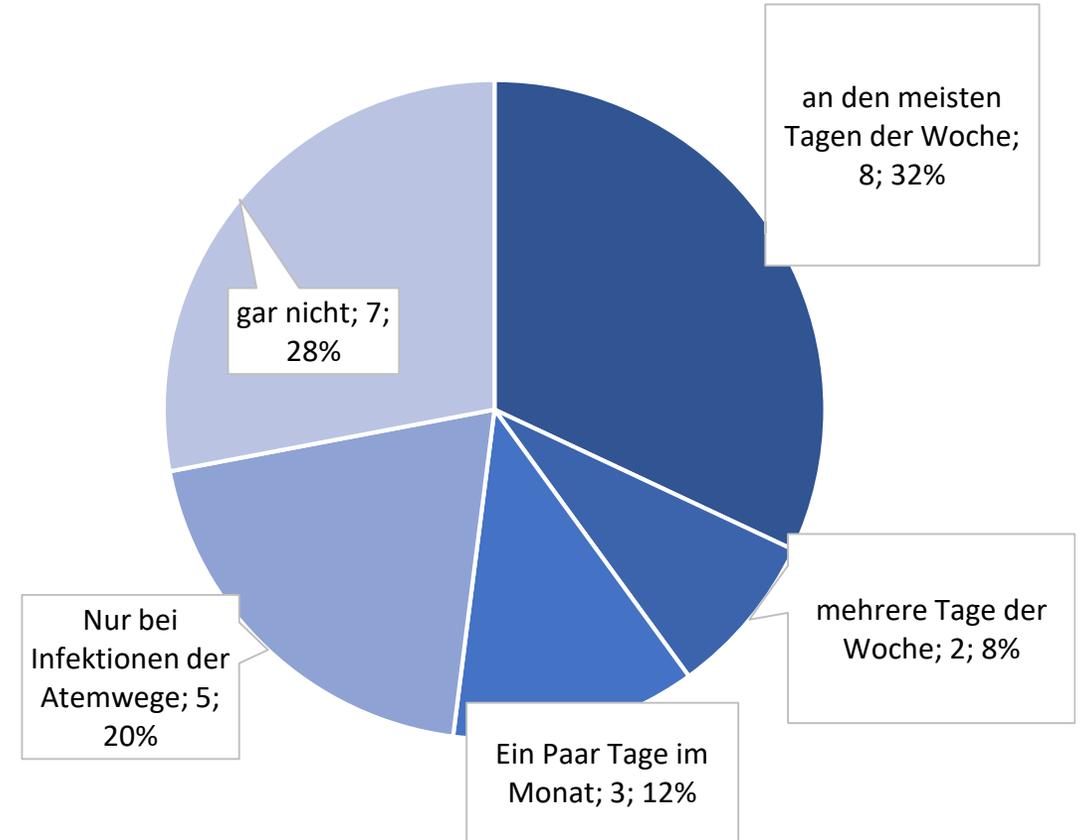
In den letzten 3 Monaten

Husten



(n=25)

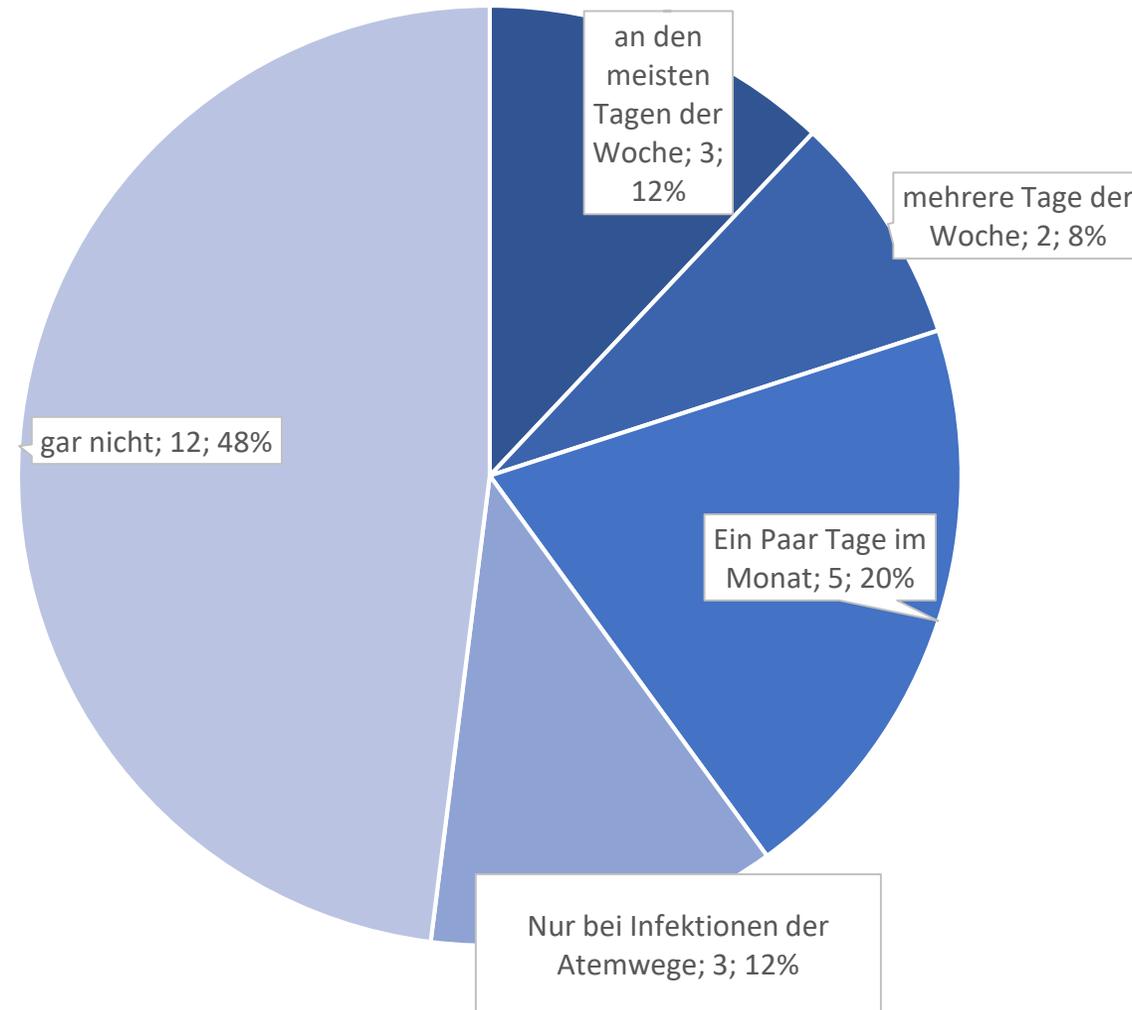
Auswurf



(n=25)

In den letzten 3 Monaten

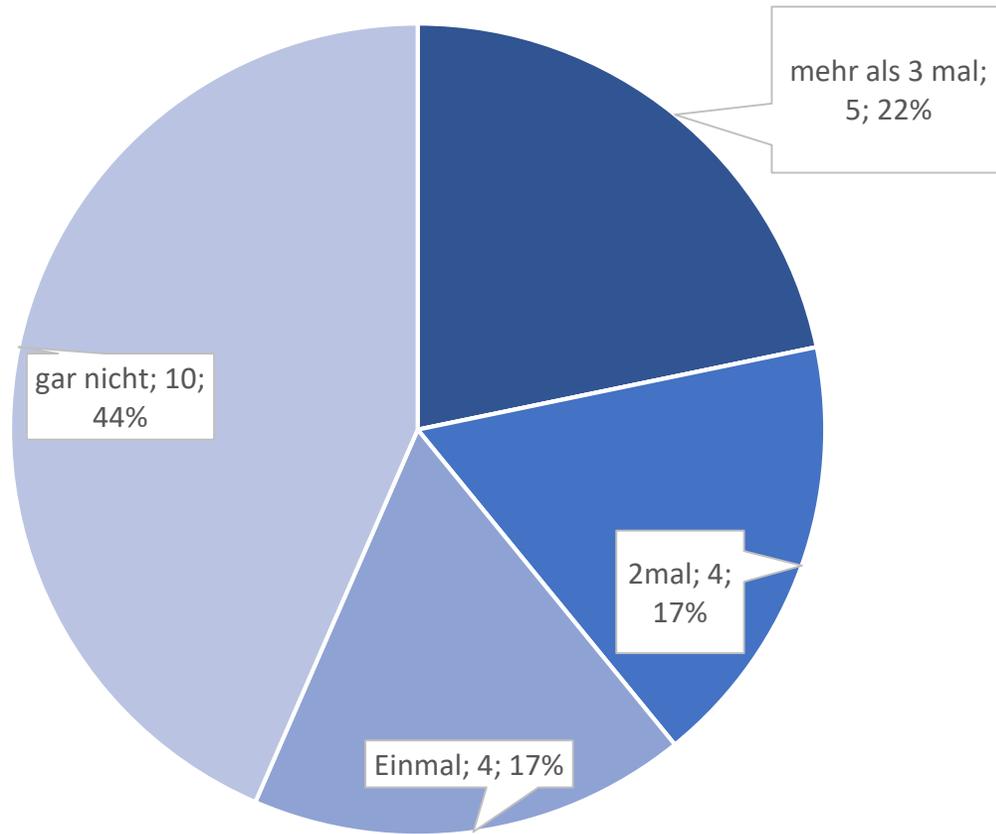
Kurzatmigkeit



(n=25)

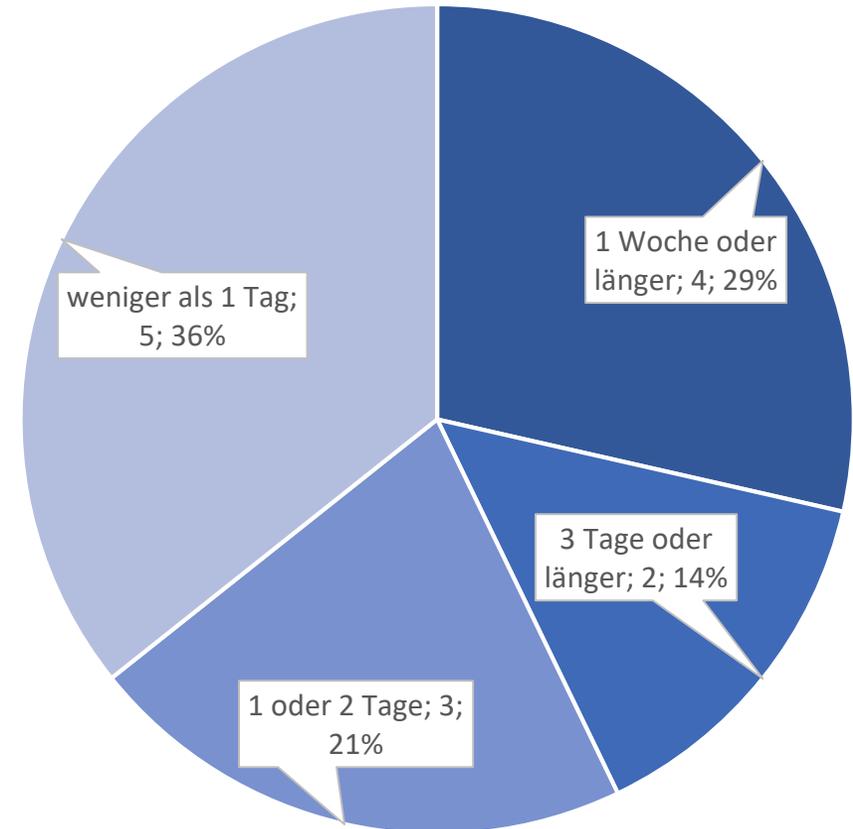
In den letzten 3 Monaten

Wie oft hatten sie Schwere oder Sehr unangenehme Atembeschwerden?



(n=23)

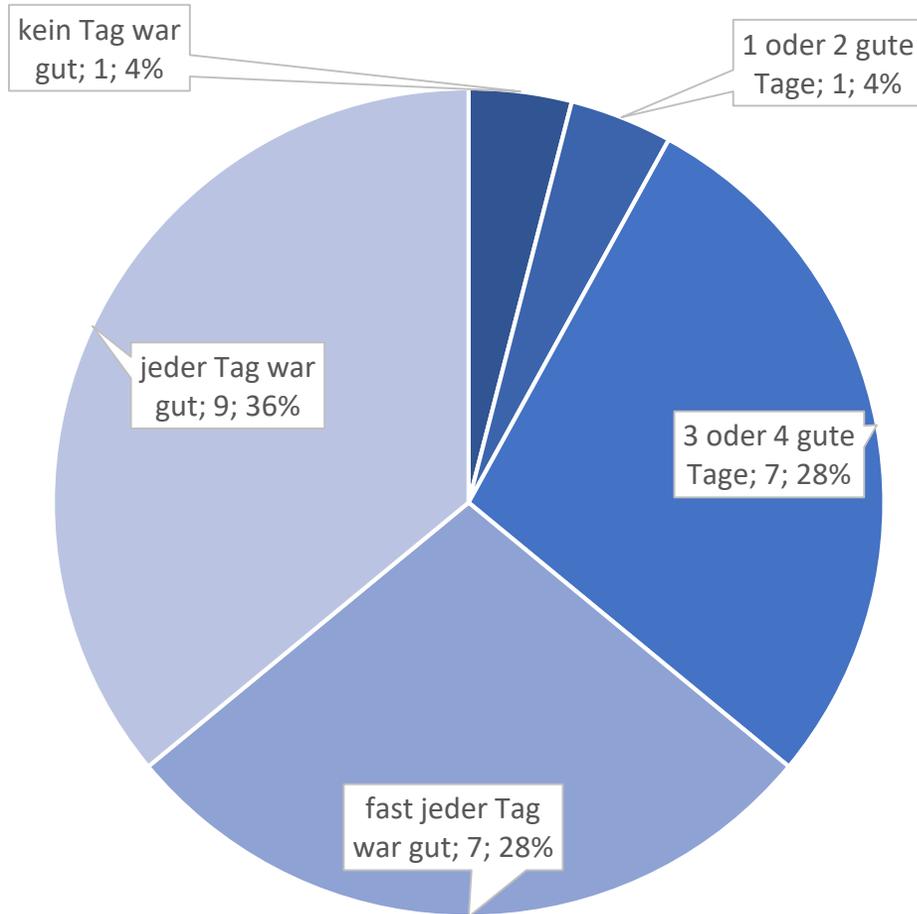
Wie lange dauerten diese schweren Atembeschwerden im schlimmsten Fall?



(n=14)

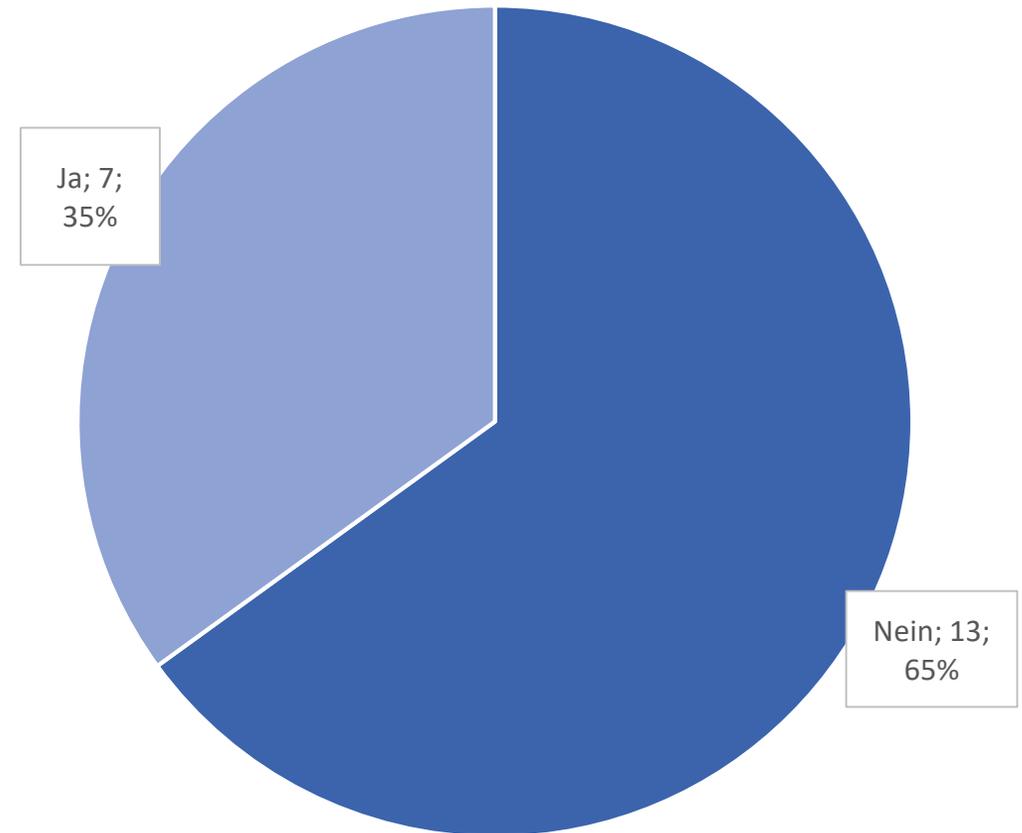
In den letzten 3 Monaten

Wie viele gute Tage hatten Sie in einer durchschnittlichen Woche?



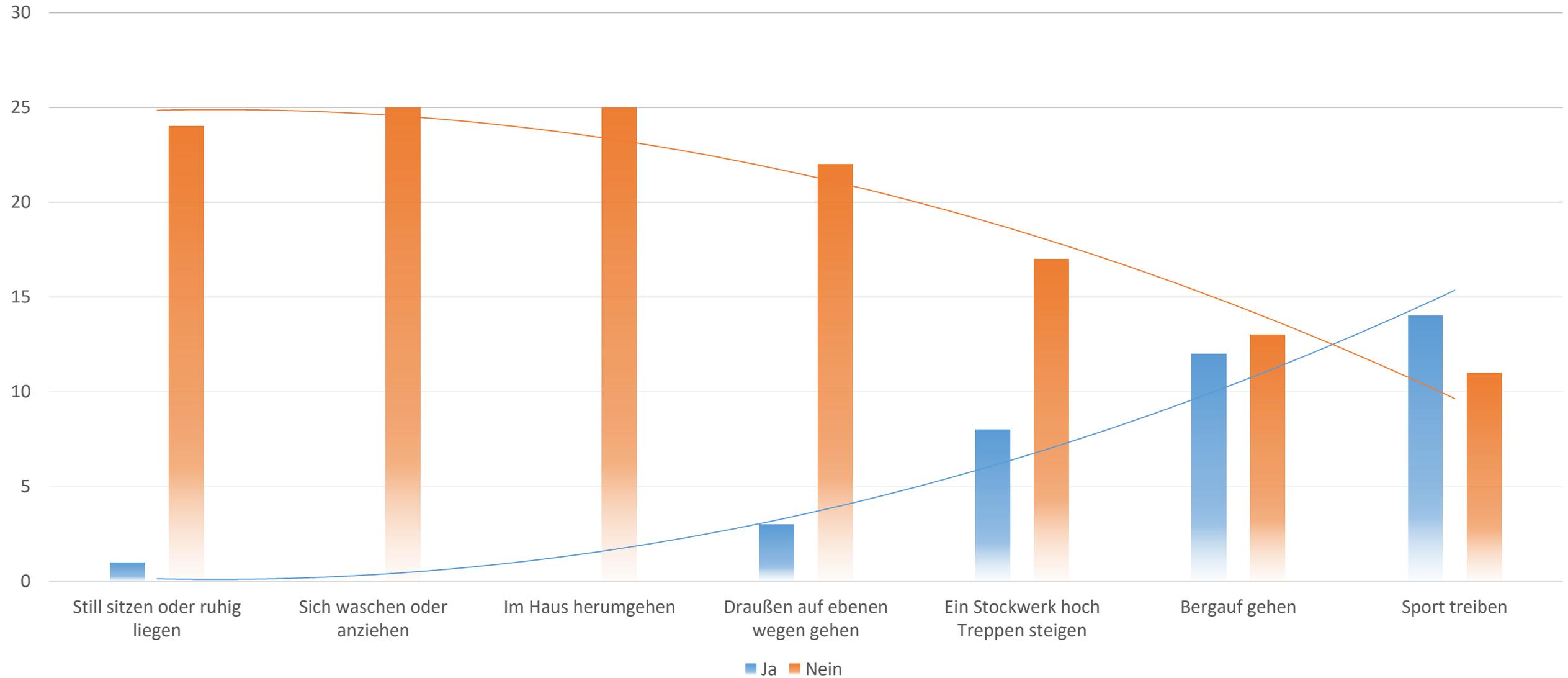
(n=25)

Wenn Sie pfeifend atmen oder keuchen, ist es morgens, nach dem Aufstehen schlimmer?



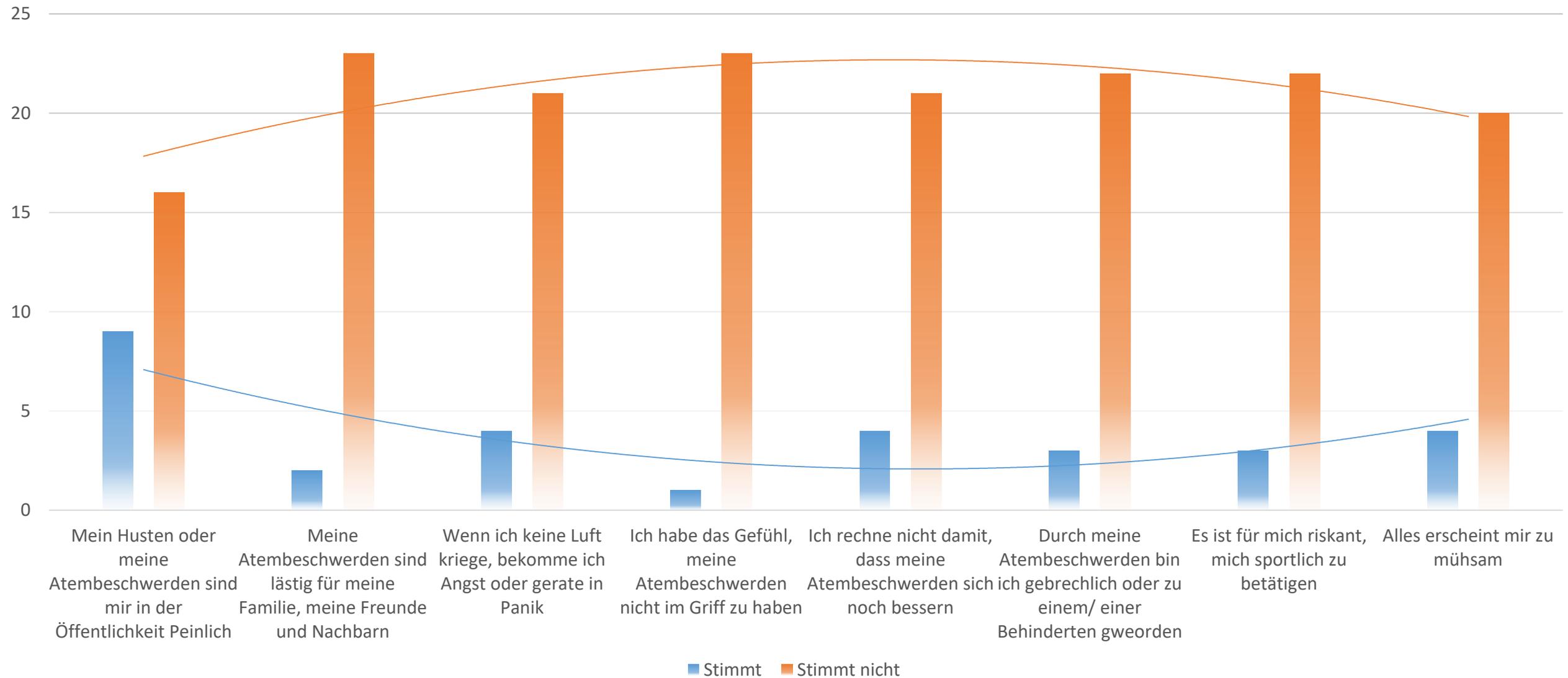
(n=20)

BEI WELCHEN TÄTIGKEITEN WERDEN SIE DERZEIT NORMALERWEISE KURZATMIG? (N=25)



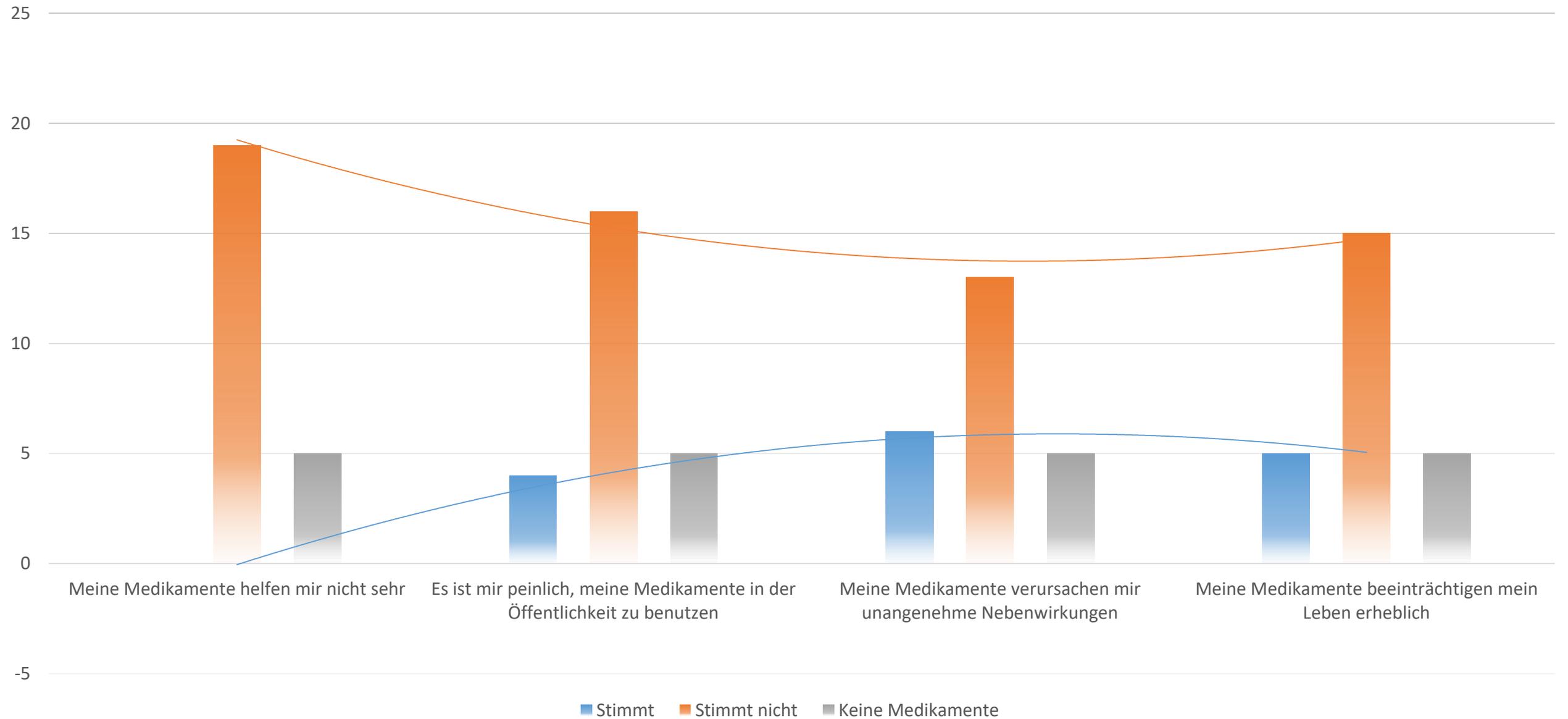
ANDERE AUSWIRKUNGEN, DIE IHRE ATEMBESCHWERDEN DERZEIT AUF SIE HABEN KÖNNEN

(N=25)

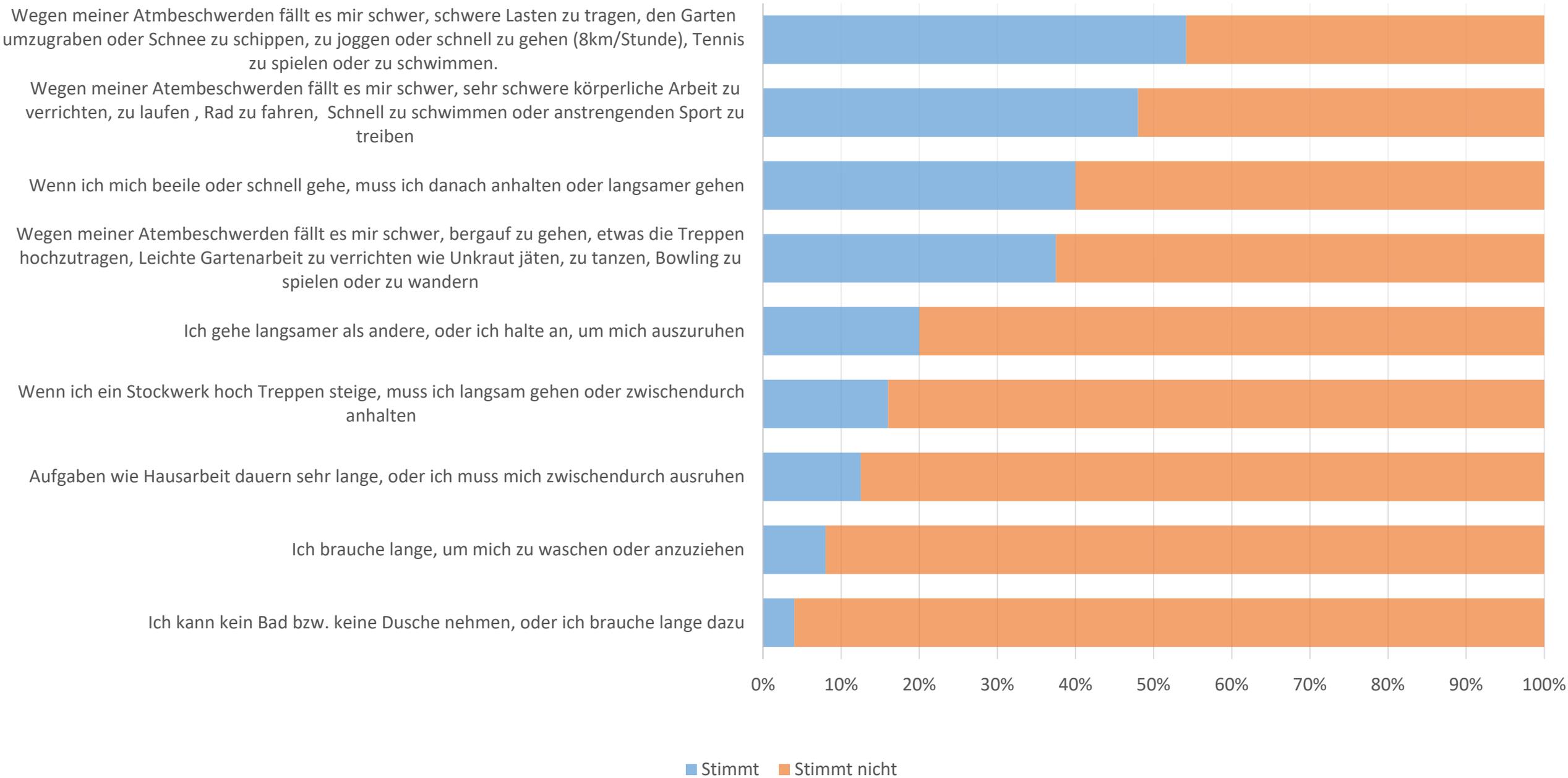


FRAGEN ZU DEN MEDIKAMENTEN

(N=25)



MÖGLICHE AUSWIRKUNG VON IHREM ATEMBESCHWERDEN AUF IHRE AKTIVITÄTEN (N=25)

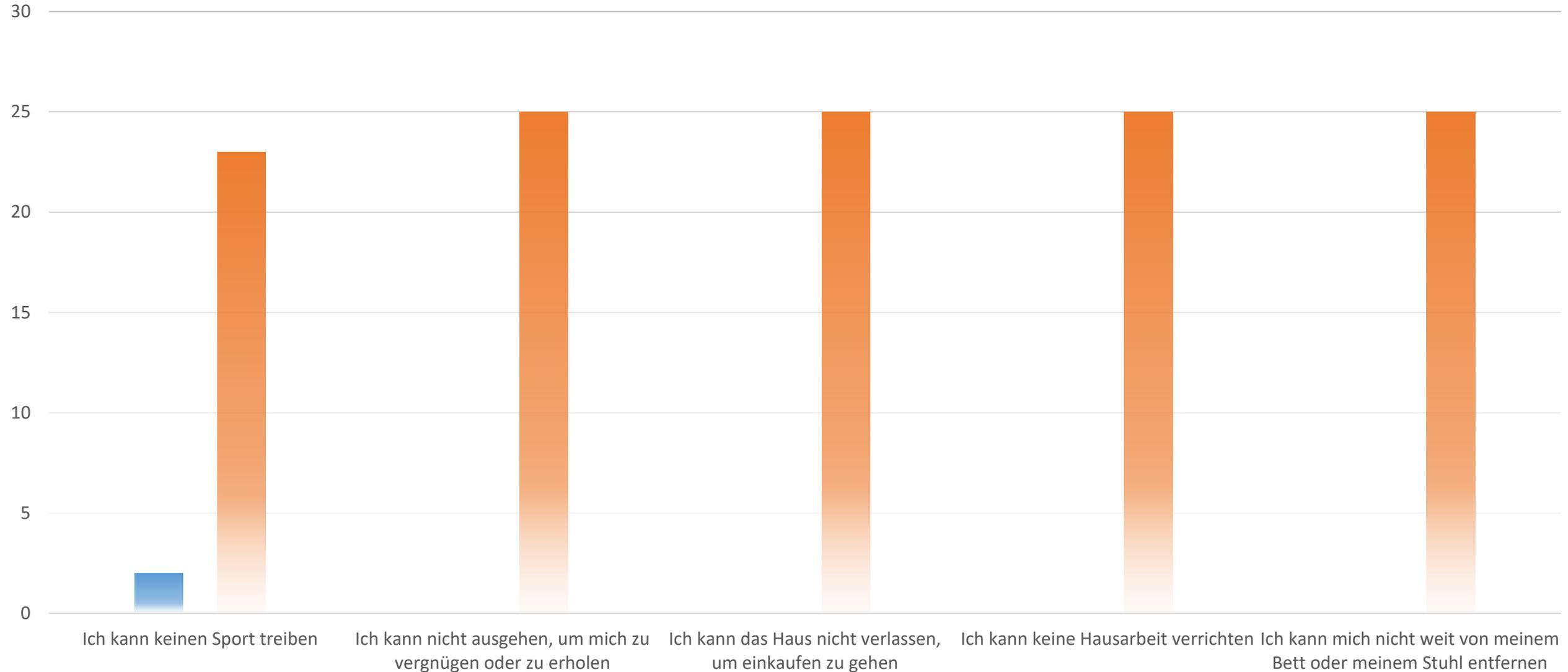


EINFLUSS VON ATEMBESCHWERDEN AUF IHR TÄGLICHES LEBEN

(N=25)

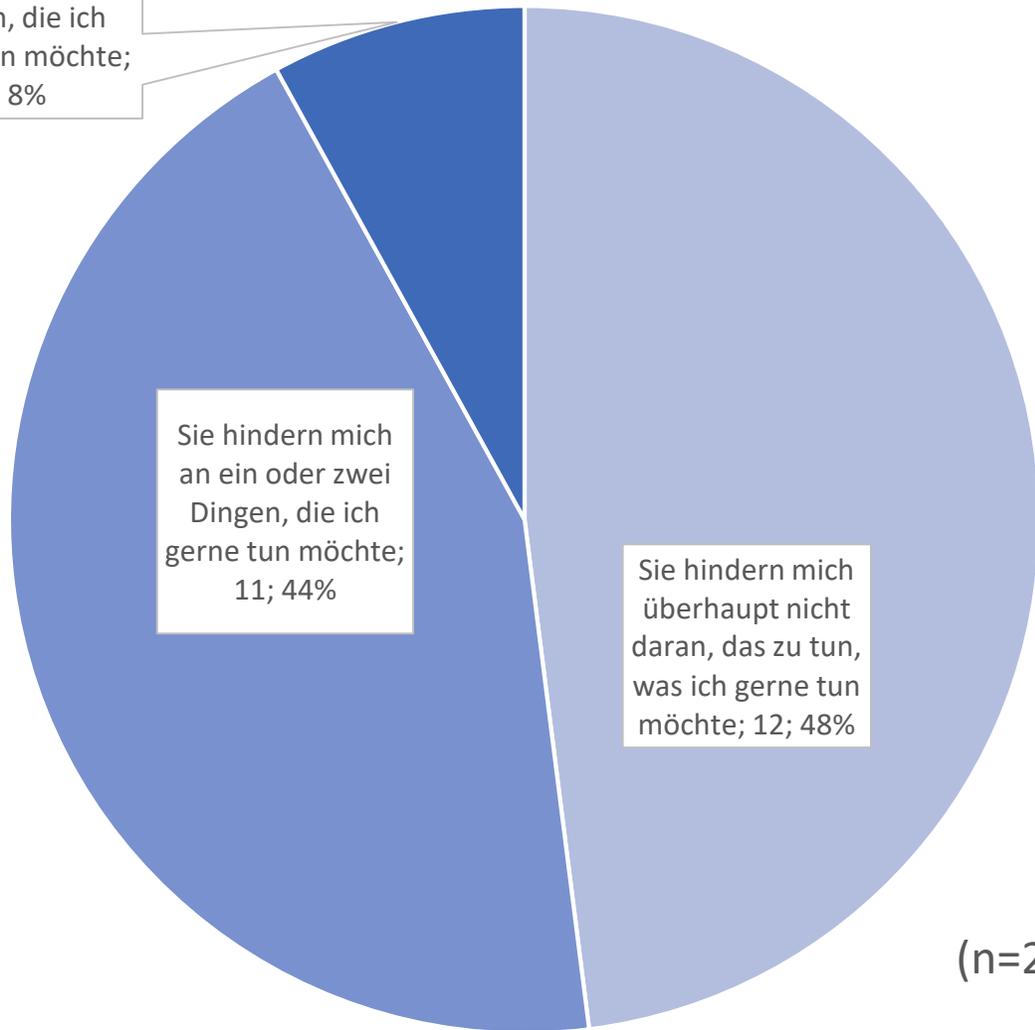
Stimmt

Stimmt nicht



Atembeschwerden

Sie hindern mich an den meisten Dingen, die ich gerne tun möchte; 2; 8%



Sie hindern mich an ein oder zwei Dingen, die ich gerne tun möchte; 11; 44%

Sie hindern mich überhaupt nicht daran, das zu tun, was ich gerne tun möchte; 12; 48%

(n=25)

Here is a list of other activities that your chest trouble may prevent you doing. (You do not have to tick these, they are just to remind you of ways in which your breathlessness may affect you):

- Going for walks or walking the dog
- Doing things at home or in the garden
- Sexual intercourse
- Going out to church, pub, club or place of entertainment
- Going out in bad weather or into smoky rooms
- Visiting family or friends or playing with children

I

Please write in any other important activities that your chest trouble may stop you doing:

Climbing, endurance sports, professional sports

Now would you tick in the box (one only) which you think best describes how your chest affects you:

- It does not stop me doing anything I would like to do 3
- It stops me doing one or two things I would like to do 3
- It stops me doing most of the things I would like to do
- It stops me doing everything I would like to do

Thank you for filling in this questionnaire. Before you finish would you please check to see that you have answered all the questions.

FAZIT

- STAT3-LOF-Hyper-IgE/Job-Syndrom-Patienten haben eine reduzierte Lebensqualität.
- Die Beeinträchtigung scheint jedoch geringer zu sein als erwartet.
- Möglicherweise erfassen die aktuellen Fragebögen zur Lebensqualität die Beeinträchtigungen/Bedürfnisse der Patienten nicht?
- Möglicherweise haben die Patienten andere Erwartungen an ihr Leben als Menschen, die nicht mit einer angeborenen Immunitätsstörung geboren wurden?
- Für die behandelnden Ärzte wird es sehr schwierig sein, Patienten für oder gegen eine Knochenmarktransplantation zu raten.

Acknowledgements

