



# Guideline parental survey & parental stress

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**umcg**



- Guideline parental survey results
- Parental stress
- Living loss
- Current parental perspective project



## Parental perspective:

- Helps researchers focus on relevant issues;
- Parents are experts on the day-to-day needs;
- Provides recommendations that are better tailored to individual needs;
- Supports clinical decision making.

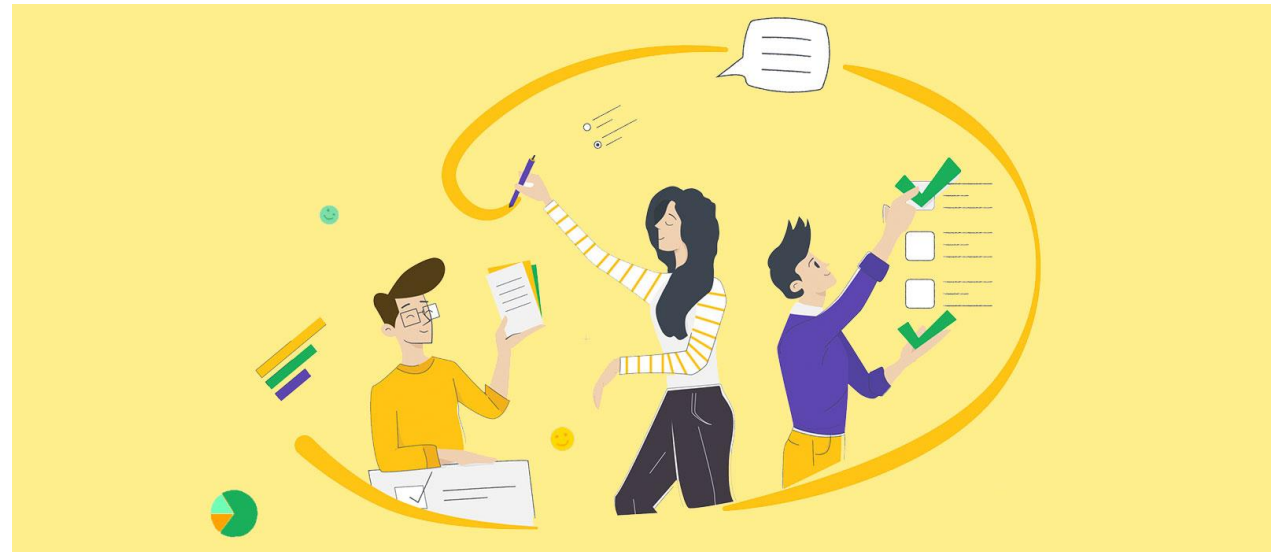


-Murad MH, Montori VM, Guyatt GH. Incorporating patient preferences in evidence-based medicine. JAMA. 2008; 300: 2483-2484

The guideline consortium developed a parental survey on:

- Somatic issues
- Genetic information
- Mental health issues
- Care needs
- Parental stress

In 10 (!) languages





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## Parental perspectives on Phelan-McDermid syndrome: Results of a worldwide survey

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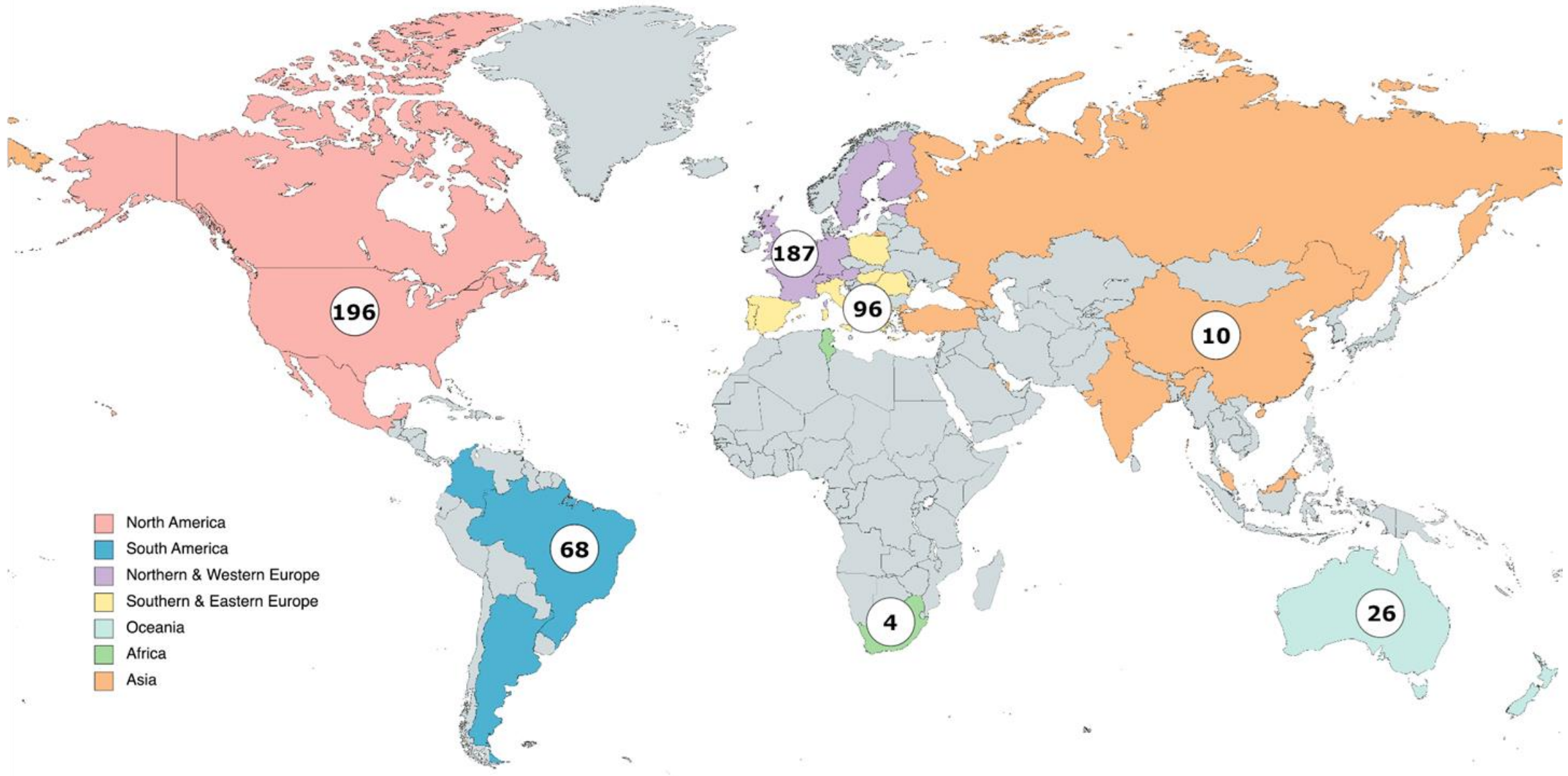
Phelan-McDermid-Gesellschaft e.V.  
Deutschland - Österreich - Schweiz



Phelan-McDermid Syndrome Foundation UK









**Table 2**  
Problems and symptoms reported in individuals with PMS.

(Somatic) problem/symptom	Prevalence, 22q13.3 deletions (n = 377, 2 missing)	Prevalence 22q13.3 deletions Schön et al.	Prevalence <i>SHANK3</i> variants (n = 107)	Prevalence, <i>SHANK3</i> variants Schön et al.	Prevalence all genotypes (n = 584, 3 missing)	Chi <sup>2</sup> p-value
<b>Development</b>						
Problems with speech and communication	97%	88%	94%	70%	97%	n.r.
Learning difficulties or intellectual disability	94%	98%	96%	96%	95%	n.r.
<b>Neurology</b>						
Problems with fine motor skills (including hand movements)	84%	n.r.	79%	n.r.	83%	n.r.
Low muscle tone (hypotonia)	81%*	74%	66%*	82%	77%	0.002*
Problems with gross motor skills (such as clumsy walking)	77%*	n.r.	63%*	n.r.	74%	0.003*
Epilepsy	25%	27%	33%	26%	25%	n.s.
<b>Senses</b>						
Low pain perception/high pain threshold	76%	65%	82%	79%	78%	n.s.
Problems with regulating body temperature	49%	37%	49%	8%	47%	n.s.
Problems with eyes and vision	30%	22%/8%	24%	26/10%	28%	n.s.
<b>Gastrointestinal problems</b>						
Constipation	46%	n.r.	41%	n.r.	48%	n.r.
Swallowing difficulties	33%	n.r.	33%	n.r.	33%	n.r.
Problems with teeth	31%	37%	24%	34%	30%	n.s.
Vomiting	17%	25% GERD	15%	17% GERD	16%	n.s.
<b>Other</b>						
Behavioural problems (generally)	70%	n.r.	78%	n.r.	71%	n.r.
Sleeping problems	57%	26%	64%	52%	59%	n.s.
Problems with feet	37%*		20%*		34%	0.001*
Problems with the kidneys and urinary tract	17%*	15%	9%*	0%	16%	0.048*
Heart problems	9%*	13%	2%*	7%	7%	0.015*
Lymphoedema	13%* (n = 371)	11%	3%* (n = 101)	0%	11%	0.004*
<b>Behavioural problems (Median age 12, 0–60)</b>						
Problems with attention and concentration	82%	57% (ASD)	80%	79% (ASD)	80%	n.s.
Problems with flexibility and adapting to changes	52%	57% (ASD)	45%	79% (ASD)	50%	n.s.
Hyperactivity	44%	29%	49%	72%	46%	n.s.
Anxious behaviour	37%*	n.r.	54%*	n.r.	41%	0.001*
Obsessions	33%	n.r.	42%	n.r.	35%	n.r.
Aggression	18%*	19%	34%*	37%	22%	0.001*
Self-injurious behaviour	16%	13%	19%	30%	17%	n.s.
Depressed behaviour	12%	n.r.	18%	n.r.	12%	n.r.
Other behavioural problems	10%	n.r.	15%	n.r.	12%	n.r.
No behavioural problems	10%	n.r.	8%	n.r.	10%	n.r.

Note: n, sample size; \* = variables that show a significant difference with  $p < 0.005$  according to Chi<sup>2</sup>, / = separate problems; GERD = gastroesophageal reflux disease; ASD = Autism Spectrum Disorder; n.r. = not reported; n.s. = not significant.

## Parental perspectives on Phelan-McDermid syndrome: Results of a worldwide survey

### Survey quotes parents:

*"I feel lost. She is becoming a teen and now everything seems to get harder".*

*"I find that the hardest and scariest part, who will take care of my adult child who will most likely always be a child".*

*"How to manage all of my child's needs without burning out"?*

**Table 4**

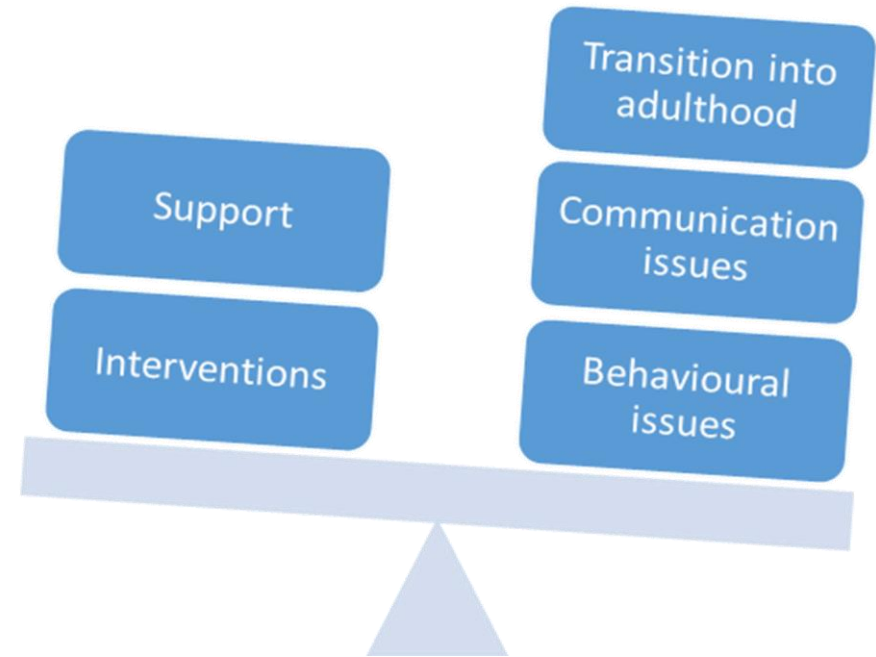
Mean scores (0–3 Likert scale) and percentage extremely stressful scores on items GSSS (n = 507) and extra added item 15.

	Topic item	Mean (0–3 Likert scale)	Percentage (%) "extremely stressful"
1.10	A genetic diagnosis causing tension within the immediate and extended family	0.94	11.1
2.	People staring when I go out in public with my child	1.12	9.9
3.	Having to make extensive preparations for my child before leaving the house	1.39	16.2
4.	Having to explain my child's condition to new people I meet	1.42	13.8
5.	Sleep deprivation, due to my child's sleeping patterns	1.58	29.8
6.	Getting my child's complex needs met through social services	1.81	30.5
7.	Not having access to professionals who have knowledge about child's condition	1.82	32.7
8.	Going to see professionals who are not knowledgeable about my child's genetic syndrome	1.93	36.6
9.	An educational placement that does not meet all of my child's needs	1.93	37.3
10.	The large amount of effort required to help my child reach developmental milestones (e.g. sitting up, self-feeding)	1.94	33.2
11.	Not being able to fully relax at home, as I need to attend to my child 24 h a day	1.96	36.5
12.	Having to be constantly vigilant about my child's state of health in case of a sudden change	2.03	39.7
13.	Arranging care (e.g. babysitting, respite) that is suitable for my child	2.08	43.6
14.	Not knowing what is bothering my child due to limited communication possibilities	2.42	61.2
15.	Worrying about the future for my child because of the lack of specialist services once they reach adulthood	2.56	68.1



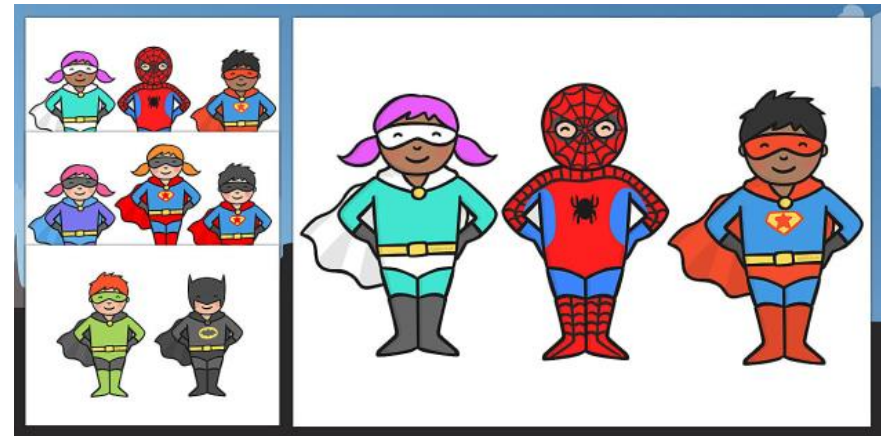
- Rare syndrome
- High parental stress
- Somatic issues
- Mental health issues
- Regression
- Sleep issues

## Parental stress balance



- Being a parent can be stressful
- Being parent of a child with a condition can be very stressful
- Being a parent of a child with a rare genetic syndrome, somatic issues, mental health issues and developmental problems.....

Has to have superpowers...



And may cause feelings of loss, being overwhelmed, stressed out. That's ok, it is normal and it will not always be like this.

# Living loss

- 'Living loss' is the term introduced by Manu Keirse to indicate the life-long mourning that you can experience when a loved one experiences a chronic disease;
- Living loss can come up any moment, expectedly or unexpectedly;
- You and your family have to adjust your life and expectations again and again, over and over;
- The grief/loss does not end, it may change however.





- You continuously have to cope with loss, it takes hard work and time to cope (it is like an extra job);
- Recognition of the loss and allowing it to be here, now and in the future, may give you space;
- People around you can best support you by listening and to recognize that these feelings of loss are there and will be there;
- This might be easier with other parents that have been there or also are there, maybe through patient organizations;
- It might also make you assertive or give strength...

Maybe even superpowers!



What do you need as a parent to be able to use your powers?



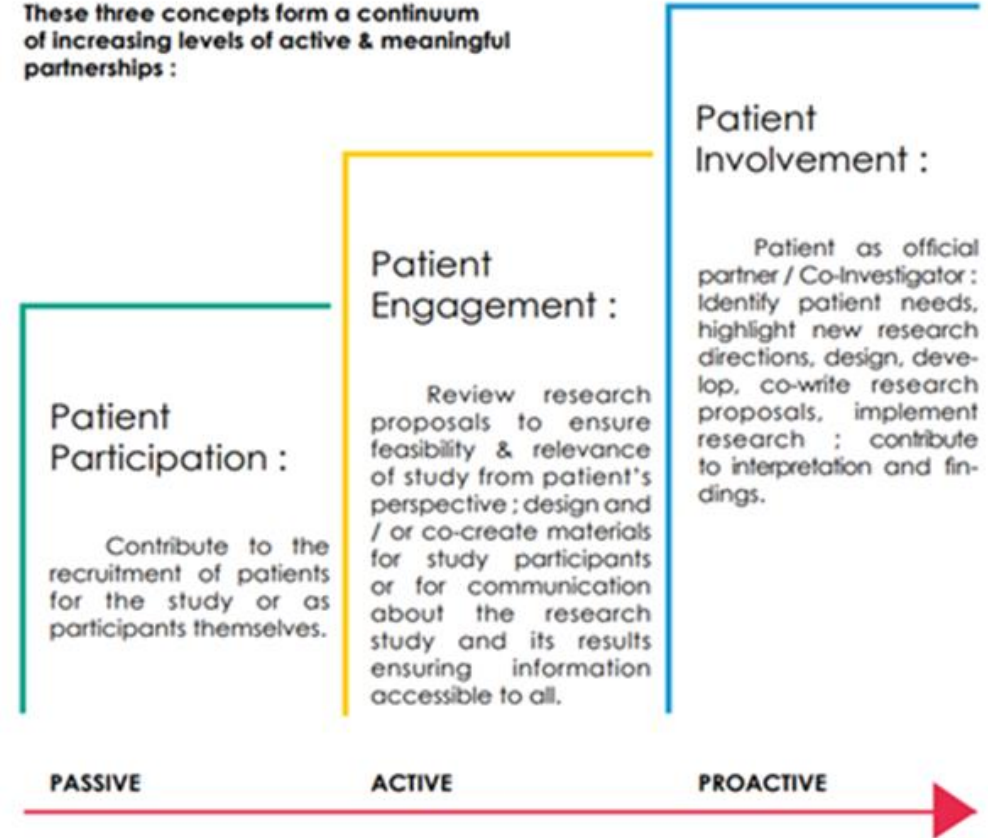


# Current parental perspective project

Survey the sequel on parents perspective on research:

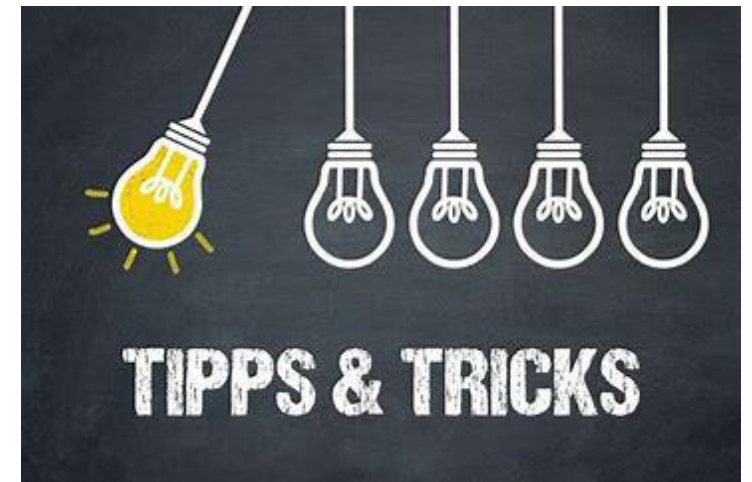
- Questions on research and innovation focus (forms of active research participation, experiences in participation);
- Which domains and topics are important to you as a parent;
- Perspective on research collaboration.

These three concepts form a continuum of increasing levels of active & meaningful partnerships :



# Possible future project??

- Third survey on tips & tricks from parents to parents
- What was truly helpful?
- Sleep, identity, learning, communication etc.
  
- Best parent practices in day-to-day life



# South African disability movement:

“NOTHING ABOUT US  
WITHOUT US”

