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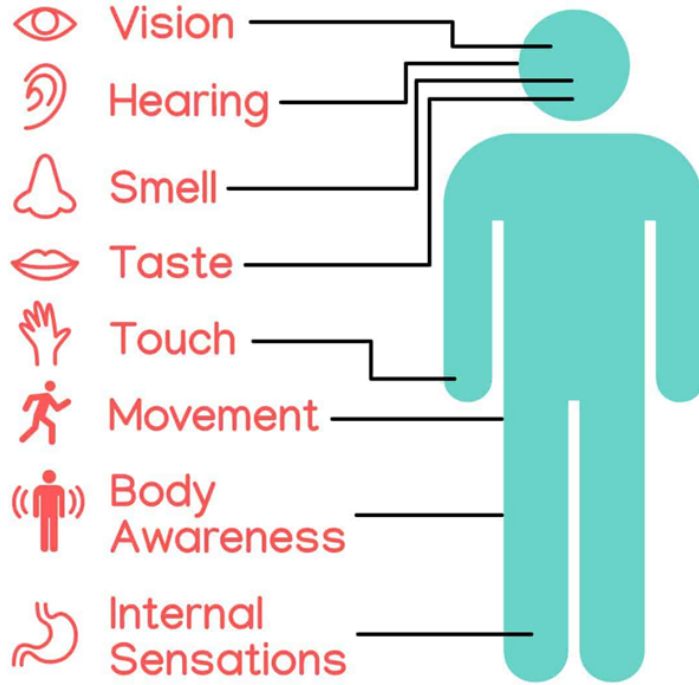
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Consensus recommendations on altered sensory functioning in Phelan-McDermid syndrome

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Senses and sensory domains



Domains:

- Visual
- Auditory
- Olfactory
- Gustatory
- Tactile
- Vestibular
- Proprioception

Sensory processing is

the neurological process of collaboration and interpretation of sensory stimuli.



TOUCH



SMELL



VISION



TASTE



HEARING

When sensory input is not leading to appropriate responses, it's called:

Sensory dysfunction or sensory processing disorder

Sensory processing

Unusual sensory responses are a criterium of autism spectrum disorder (DSM-5):
hyper- or hyporeactivity to sensory input or sensory seeking behaviour

- Hyperreactivity: a strong reaction to sensory stimuli
- Hyporeactivity: delayed/absent response
- Sensory seeking : fascination with certain sensory stimuli

Sensory functioning in PMS

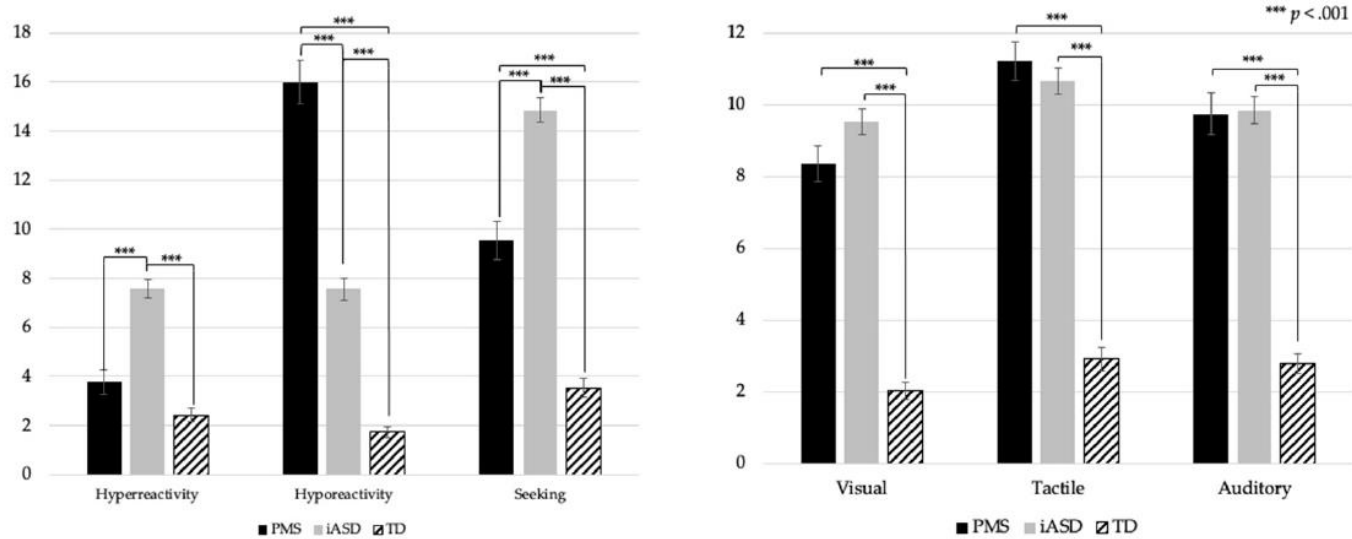






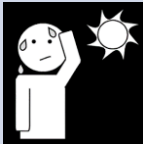

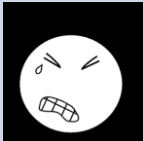
Figure 1. SAND domain (Hyperreactivity, Hyporeactivity, Seeking) and modality (Visual, Tactile, Auditory) mean scores for PMS, iASD, and TD groups. Tavassoli et al. Sensory Reactivity phenotype in Phelan–McDermid Syndrome Is Distinct from Idiopathic ASD. *Genes* **2021**

More hypo-reactivity symptoms in visual and particularly auditory domain and a sensitivity to touch, compared to people with ASS/TD.

Domains

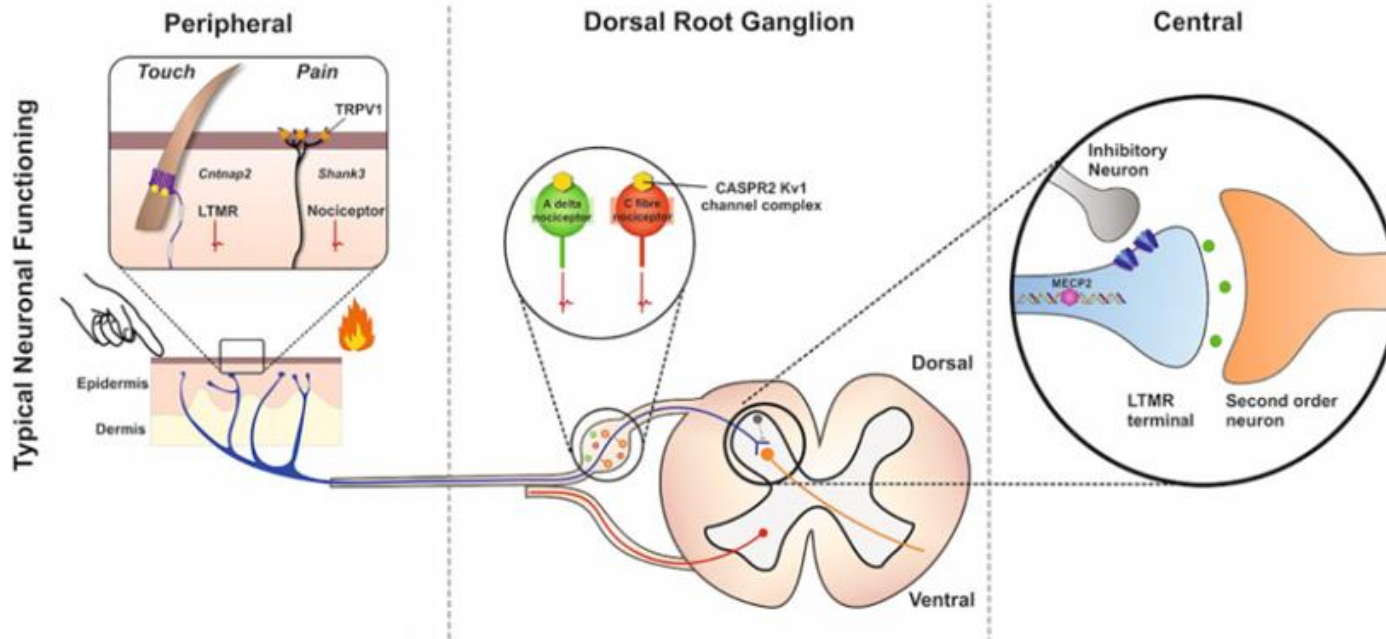
Domain	Medical problems	Sensory processing features
Proprioception & vestibular 	Ring chromosome 22; possibility of a tumor causing dizziness /balance problems	Sometimes stimulus seeking: lying on the floor or moving on the knees
Hearing 	Mostly normal. Ring chromosome 22; possibility of a tumor causing hearing loss	Delayed response to verbal and auditory cues Sometimes difficulties to distinguish words from background noises
Vision 	Vision disturbances (refraction, strabismus) in 22% -29%	Delayed or absent responses to the sight
Smell /taste 	None known	Sometimes stimulus seeking: licking objects, smelling things or people

Tactile domain

		Medical /sensory problems
Heat regulation		Overheating or turning red easily Decreased perspiration: *22q13.3 deletion in 37% *SHANK3 variants in 8%
Tactile sensibility		Hypersensitivity to touch in 46%
Experience of pain		Reduced pain response: *22q13.3 deletion in 65% *SHANK3 variants in 79% increase with age from 69% at 5 years to 89% in adults

What is the mechanism behind it?

SHANK3 protein plays a role in the nerve cells of the pain nerves



➔ Underlying mechanism remains unclear

To assess the sensory profile:

Assessing should be done by a trained person

(f.e. occupational or physical therapist, psychologist or behavioural scientist).

Validated instruments :

- The Short Sensory Profile (SSP), shortened form of:
 - Dunn's Sensory Profile 2 caregiver questionnaire
 - The SAND (Sensory Assessment for Neurodevelopmental Disorders)
- Adolescent/Adult Sensory Profile (AASP)

After assessing the sensory profile, appropriate advise can be given



Examples



To assess pain, use validated instruments

Non-communicating Children's Pain Checklist – Revised (NCCPC-R)

NAME: _____ UNIT FILE #: _____ DATE: _____ (dd/mm/yy)
 OBSERVER: _____ START TIME: _____ AM/PM STOP TIME: _____ AM/PM

How often has this child shown these behaviours in the last 2 hours? Please circle a number for each item. If an item does not apply to this child (for example, this child does not eat solid food or cannot reach with his/her hands), then indicate "not applicable" for that item.

0 = NOT AT ALL 1 = JUST A LITTLE 2 = FAIRLY OFTEN 3 = VERY OFTEN NA = NOT APPLICABLE

I. Vocal					
1. Moaning, whimpering, whimpering (fairly soft)	0	1	2	3	NA
2. Crying (moderately loud)	0	1	2	3	NA
3. Screaming/yelling (very loud)	0	1	2	3	NA
4. A specific sound or word for pain (e.g., a word, cry or type of laugh)	0	1	2	3	NA

II. Social					
5. Not cooperating, cranky, irritable, unhappy	0	1	2	3	NA
6. Less interaction with others, withdrawn	0	1	2	3	NA
7. Seeking comfort or physical closeness	0	1	2	3	NA
8. Being difficult to distract, not able to satisfy or pacify	0	1	2	3	NA

III. Facial					
9. A furrowed brow	0	1	2	3	NA
10. A change in eyes, including: squinting of eyes, eyes opened wide, eyes frowning	0	1	2	3	NA
11. Turning down of mouth, not smiling	0	1	2	3	NA
12. Lips puckering up, tight, pouting, or quivering	0	1	2	3	NA
13. Clenching or grinding teeth, chewing or thrusting tongue out	0	1	2	3	NA

IV. Activity					
14. Not moving, less active, quiet	0	1	2	3	NA
15. Jumping around, agitated, fidgety	0	1	2	3	NA

V. Body and Limbs					
16. Flaccid	0	1	2	3	NA
17. Self-spastic, tense, rigid	0	1	2	3	NA
18. Gesturing to or touching part of the body that hurts	0	1	2	3	NA
19. Protecting, favoring or guarding part of the body that hurts	0	1	2	3	NA
20. Flinching or moving the body part away, being sensitive to touch	0	1	2	3	NA
21. Moving the body in a specific way to show pain (e.g. head back, arms down, curls up, etc.)	0	1	2	3	NA

VI. Physiological					
22. Shivering	0	1	2	3	NA
23. Change in color, pallor	0	1	2	3	NA
24. Sweating, perspiring	0	1	2	3	NA
25. Tears	0	1	2	3	NA
26. Sharp intake of breath, gasping	0	1	2	3	NA
27. Breath holding	0	1	2	3	NA

VII. Eating/Sleeping					
28. Eating less, not interested in food	0	1	2	3	NA
29. Increase in sleep	0	1	2	3	NA
30. Decrease in sleep	0	1	2	3	NA

SCORE SUMMARY:	I	II	III	IV	V	VI	VII	TOTAL
Category:								
Score:								

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Paediatric Pain Profile

Baseline assessments

Pain Profile

Most troublesome pain (Pain A)

- For each item please circle the number that best describes your child's behaviour when they have this pain.
- Enter the number you have circled in to the "score" column.
- Add up the numbers in the "score" column to give the total score.
- Record the score on the Summary Graph

When my child has this pain, he or she...	Not at all	A little	Quite a lot	A great deal	Score
Is cheerful	3	2	1	0	
Is sociable or responsive	3	2	1	0	
Appears withdrawn or depressed	0	1	2	3	
Cries / moans / groans / screams or whimpers	0	1	2	3	
Is hard to console or comfort	0	1	2	3	
Self-harms e.g. biting self or banging head	0	1	2	3	
Is reluctant to eat / difficult to feed	0	1	2	3	
Has disturbed sleep	0	1	2	3	
Grimaces / screws up face / screws up eyes	0	1	2	3	
Frowns / has furrowed brow / looks worried	0	1	2	3	
Looks frightened (with eyes wide open)	0	1	2	3	
Grinds teeth or makes mouthing movements	0	1	2	3	
Is restless / agitated or distressed	0	1	2	3	
Tenses / stiffens or spasms	0	1	2	3	
Flexes inwards or draws legs up towards chest	0	1	2	3	
Tends to touch or rub particular areas	0	1	2	3	
Resists being moved	0	1	2	3	
Pulls away or flinches when touched	0	1	2	3	
Twists and turns / tosses head / writhes or arches back	0	1	2	3	
Has involuntary or stereotypical movements / is jumpy / startles or has seizures	0	1	2	3	
					TOTAL

Please tick the box next to the word that best describes the severity of this pain

None Mild Moderate Severe Very severe

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Chronic Pain Scale for Nonverbal Adults With Intellectual Disabilities (CPS-NAID)

Please indicate how often this person has shown the signs referred to in items 1-24 in the last 5 minutes. Please circle a number for each item. If an item does not apply to this person (for example, this person cannot reach with his/her hands), then indicate "not applicable" for that item.

0 =	Not present at all during the observation period. (Note: if the item is not present because the person is not capable of performing that act, it should be scored as "NA").
1 =	Seen or heard rarely (hardly at all), but is present.
2 =	Seen or heard a number of times, but not continuous (not all the time).
3 =	Seen or heard often, almost continuous (almost all the time), anyone would easily notice this if they saw the person for a few moments during the observation time.
NA =	Not applicable. This person is not capable of performing this action.

0 = Not at all	1 = Just a little	2 = Fairly Often	3 = Very Often	NA = Not Applicable	
1. Moaning, whining, whimpering (fairly soft)	0	1	2	3	NA
2. Crying (moderately loud)	0	1	2	3	NA
3. A specific sound or word for pain (e.g. A word, cry or type of laugh)	0	1	2	3	NA
4. Not cooperating, irritable, unhappy	0	1	2	3	NA
5. Less interaction with others, withdrawn	0	1	2	3	NA
6. Seeking comfort of physical closeness	0	1	2	3	NA
7. Being difficult to distract, not able to satisfy or pacify	0	1	2	3	NA
8. A furrowed brow	0	1	2	3	NA
9. A change in eyes, including: squinting of eyes opened wide, eyes frowning	0	1	2	3	NA
10. Turning down of mouth, not smiling	0	1	2	3	NA
11. Lips puckering up, tight, pouting or quivering	0	1	2	3	NA
12. Clenching or grinding teeth, chewing or thrusting tongue out	0	1	2	3	NA
13. Not moving, less active, quiet	0	1	2	3	NA
14. Stiff, spastic, tense, rigid	0	1	2	3	NA
15. Gesturing to or touching part of the body that hurts	0	1	2	3	NA
16. Protecting, favoring or guarding part of body that hurts	0	1	2	3	NA
17. Flinching or moving the body part away, being sensitive to touch	0	1	2	3	NA
18. Moving the body in a specific way to show pain (e.g. Head back, arms down, curls up, etc.)	0	1	2	3	NA
19. Shivering	0	1	2	3	NA
20. Change in colour, pallor	0	1	2	3	NA
21. Sweating, perspiring	0	1	2	3	NA
22. Tears	0	1	2	3	NA
23. Sharp intake of breath, gasping	0	1	2	3	NA
24. Breath holding	0	1	2	3	NA

Subtotals:

1. For each subtotal write the number of times each value was chosen	NA	1x	2x	3x	NA
2. Multiply the value of each selection by how many times that value was chosen		=	=	=	Total:
3. Add each subtotal to find the total score		=	=	=	

Recommendations

1.

Screen vision and hearing

2.

Assess sensory functioning

3.

Altered behaviour: assess pain and/or sensory functioning

4.

Be aware of a reduced response to pain, heat regulation or sudden sounds

Thank you for your attention !

