

# Physical Therapy

HNRNPH2 Family Meeting

26th – 27th July 2018

Center for Autism and the Developing Brain (CADB)

NewYork-Presbyterian Hospital – Weill Cornell Medical College

Discuss global developmental struggles and therapies in the areas of occupation and physical therapy.

Traditional and “Untraditional Perspectives: Helpful equipment, alternative therapies, medications & naturopathic supplements”.

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# Physical Therapy

## *Brief History*

- 4000 a.C and 395 d.C : physical agents (sun, light, heat, water and electricity), massage and physical exercises;
- 2698 a.C registration of works in kinesiotherapy;
- Middle Ages: divine plan – valorization of the soul;
- Renaissance 15th and 16th century: scientific growth and literary – maintenance of the normal state of healthy individuals;
- 18th and 19th century: industrialization, proliferation of new diseases, valorisation and prioritization of medicine;
- Physiotherapy continued to evolve with their specializations;
- Each country declares recognition of the profession in its time;

# Physical Therapy

*How can a man stand in a biped or inclined position with the wind blowing on him?*

*Charles Bell*

Since the 18th century:

- Charles Bell: posturology
- Romberg / Baron: the obvious importance of the eyes
- Longet: proprioception of paravertebral muscles
- Flourens: vestibular influence
- **Sherrington**: the synapses and the muscular sense
- Kendal: the joint of the body at a certain moment
- J. Paillard: body situated and body identified - psychophysiological approximation of the notion of body schema
- Babinski: observing the defects of coordination between posture and movement in cerebellar patients, will be to whom we owe the first information of postural adjustments associated with the voluntary movement. Babinski Reflex.
- Piaget: the theory of the stages of human development and education in a profound way. Cognition and Motor.
- ... Methods of evaluation: scales, reflexes,...

# Research

- A century ago different works carried out consider the postural and cognitive system as a “whole structured” with multiple inputs resulting in several complementary functions:
  - Fight against gravity and maintain an upright position;
  - Oppose external forces;
  - Put us in structured time space;
  - Balance in the movement, guide us and reinforce us;

To achieve these neurophysiological effects, the organism uses different sources

- The exteroceptives, place us in relation to our environment (touch, sight, hearing);
- The proprioceptors, situate the different parts of our body in relation to the whole in a certain position;
- The higher centers of the brain integrate the selectors of the strategies, the cognitive processes and treat the information of the arrival and departure of the sources;

# Therapy and Methods, which to choose?



- I could quote here a list of these;
- From the oldest to the most modern;
- Point of view: duration, frequency and repetition;
- Environment of the child: Influence their acquisitions;

# Variants in HNRNPH2 on the X Chromosome Are Associated with a Neurodevelopmental Disorder in Females

Jennifer M. Bain,<sup>1</sup> Megan T. Cho,<sup>2</sup> Aida Telegrafi,<sup>2</sup> Ashley Wilson,<sup>3</sup> Susan Brooks,<sup>4</sup> Christina Botti,<sup>4</sup> Gordon Gowans,<sup>5</sup> Leigh Anne Autullo,<sup>5</sup> Vidya Krishnamurthy,<sup>6</sup> Marcia C. Willing,<sup>7</sup> Tomi L. Toler,<sup>7</sup> Bruria Ben-Zev,<sup>8</sup> Orly Elpeleg,<sup>9</sup> Yufeng Shen,<sup>10</sup> Kyle Retterer,<sup>2</sup> Kristin G. Monaghan,<sup>2</sup> and Wendy K. Chung<sup>3,11,\*</sup>

For us: October 3rd of 2016;

Contact with the first families by virtual ways;

January 2017 New York 1st meeting;

# Return to São Paulo

- Case report regarding a female child diagnosed with the genetic variant HNRPHN2 at 10 years of age.
- This study aimed to report her path and the history of physiotherapy in her life quality between 2007 and 2017.
- For that purpose, all medical reports were analyzed, as well as laboratory tests, 77 reports of physiotherapy and other therapies to which the child was submitted in this period.
- Besides, all the results of these procedures are reported at length. The paper authored by Dr. Jennifer Bain et al. (2016)<sup>1</sup> mentions that this genetic mutation had been detected **in six girls** until the date of that publication.
- The child's diagnosis was possible only after the genetic laboratory had access to the publication, also in 2016. In the previous year, the child had undergone the complete genetic sequencing examination with negative result.



To define

Rare diseases: In search of a diagnosis, the importance of early  
physiotherapy!

# Conclusion

- The conclusion is that early physiotherapy practice while waiting for a diagnosis was extremely important for this child's progression.
- *In Louise's case, evolution and acquisitions depended on how and how often she was stimulated.*
- On the other hand, even with the sampling still small, besides these girls live in different countries and ways of life differs, the rarity and the recent discovery of this genetic mutation, the challenge is to gather as much information about the physiotherapies and the multidisciplinary follow-up adopted individually and compare the results obtained.
- With this, it will be possible to bring the formation in the light of science to collaborate with the evolution of each one of these children and of the others as yet undiagnosed.

Thank you very much

*Do not set limits we do not know.*



Louise's father, François-Paul Boutard