



annotate hpo codes from phenotypes and indicate what text is associated with each hpo code:

Telehealth on 9th June 2022 1pm (both parents)

Started off conversation with father

Mother joined at 1:07

According to father:

Resting better at home

Took alot of adjustments to get back to her routines

- Have been monitoring HR, improving, baseline now ~100bpm
- BP improved
- CIC minimal
- R hand strength improved
- Coordination improving
- Still needs some assistance with walking - still wobbly

Have not seen psychiatrist

Parents keen on cutting back some medications

Discussed about mitochondchondrial disorders - for this particular condition, a higher percentage of heteroplasmy is associated with a probability of being affected
However the percentage is not a reflection of disease severity

To continue cardiac monitoring

To continue monitoring eyes - possibility of developing retinitis pigmentosa

For respi followup, spirometry planned with Dr

Father enquired if monitoring is for intervention

- for cardiac monitoring, there are medications that can be helpful, if required
- for eye, more for monitoring the development of RP. at this time, there are no drug therapies

Menarche April 2021

Weakness noted Dec 2021

Ptosis noted Jan 2022

X is likely considered within the mild to moderate range



- HP:0000855 Abnormality of blood pressure - "BP improved"
- HP:0003324 Cerebellar ataxia - "Still needs some assistance with walking - still wobbly"
- HP:0001290 Hypertonia - "R hand strength improved"
- HP:0000962 Ptosis - "Ptosis noted Jan 2022"
- HP:0003011 Weakness - "Weakness noted Dec 2021"
- HP:0012637 Abnormality of cardiac function - "To continue cardiac monitoring"
- HP:0000486 Visual impairment - "To continue monitoring eyes - possibility of developing retinitis pigmentosa"
- HP:0011017 Mitochondrial inheritance - "Discussed about mitochondrial disorders - for this particular condition, a higher percentage of heteroplasmy is associated with a probability of being affected."
- HP:0001607 Respiratory insufficiency - "For respi followup, spirometry planned with Dr"

