CONGENITAL HYPOPITUITARISM

1. What is the pituitary and the hypothalamus?

The pituitary is a small endocrine gland that looks like a grape, it is connected to the hypothalamus, that resides right above it, with the pituitary stalk and secretes many hormones, that orchestrate body's function and homeostasis. The anterior lobe of the pituitary secretes growth hormone (GH) that stimulates growth, the thyroid stimulating hormone (TSH) controlling thyroid function, the gonadotrophins (LH and FSH) that orchestrate puberty, controlling ovaries and testes respectively, adrenocorticotropic hormone (ACTH), that controls the adrenals and cortisol production and finally prolactin. The posterior lobe secretes antidiuretic hormone (ADH) that controls the body's water balance and oxytocin that has an important role during delivery and breastfeeding. The hypothalamus functions just like a computer's processor and plays an important role in vital functions of the body like thirst and appetite, and controls hormone secretion from the pituitary gland. The pituitary gland is often called the master gland because it controls several other hormone glands in the body, including the thyroid and adrenals, the ovaries, and testes.

2. What is pituitary deficiency?

Pituitary deficiency or hypopituitarism is the state when pituitary cannot produce and secrete one or more of the pituitary hormones. It can be congenital or acquired, partial or complete.

3. What is Congenital hypopituitarism?

Congenital hypopituitarism is characterized by deficiencies or insufficiencies of one or more of the pituitary hormones such as growth hormone (GH), thyroid stimulating hormone (TSH), adrenocorticotropic hormone (ACTH), prolactin and gonadotropins (LH, FSH). Congenital means that that the person was born with it, not necessarily a heritable disorder.

4. What are the causes of Congenital Pituitary Deficiency?

Congenital pituitary deficiency occurs either due to mutations of certain genes that control pituitary cell functions or due to abnormalities of the anatomy in the pituitary region. In addition, it may be the result of injury (e.g. during a difficult birth). Nevertheless, in many cases no particular cause for the deficiencies or insufficiencies is found.

5. What are the symptoms of Congenital Pituitary Deficiency?

Symptoms vary, depending on the type and the severity of pituitary hormone deficiencies. Depending on the affected hormonal axis the following may occur:

- Growth hormone deficiency (GHD): Growth failure and short stature, hypoglycemia, relative obesity.
- Secondary hypothyroidism (TSH deficiency): constipation, poor growth, hair loss, weight gain)

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- Secondary adrenal deficiency (ACTH deficiency): ACTH deficiency can lead to secondary adrenal insufficiency, which means insufficient production of cortisol from the adrenal glands. Patients may have symptoms such us: fatigue, episodes of vomiting and hypoglycaemia. Cortisol is the hormone responsible for the body's response to stress, hence when it is deficient, there is a risk of the body collapsing during common situations such us an infection or trauma. The condition can be very serious, it is called adrenal crisis and needs immediate medical treatment.
- LH and FSH deficiency: pubertal failure, infertility
- Central diabetes insipidus (ADH deficiency): Polyuria, polydipsia, dehydration.

6. How is Growth hormone deficiency (GHD) diagnosed?

The diagnosis is confirmed by measuring the level of growth hormone production in response to a stimulation test, which generally requires a morning in the hospital.

7. What are the Signs and Symptoms of isolated Growth hormone deficiency (GHD)?

GHD does not affect intrauterine growth, at least not to an extent measurable by birth weight or length, but usually from around the age of two growth is slower than normal. A child with GHD will have short stature and look young for its age, however the body will be in proportion and it will have normal facial features and intelligence. However, prior to treatment children with GHD may be overweight as growth hormone also controls the level of fat under the skin.

8. What is the treatment for Congenital Pituitary Deficiency?

Treatment is given to replace the hormones of the pituitary gland or the hormones of the target organs that are insufficient.

9. What is the treatment for Growth hormone deficiency (GHD)?

GHD is treated with a daily injection of growth hormone that replaces the hormone the body is failing to produce. During treatment it is important that height is monitored regularly and sometimes a bone scan and brain scan, specifically of the hypothalamic-pituitary region, might be done. There are usually no side-effects from treatment. For most children, the treatment with growth hormone will result in the child reaching the expected height.

10. Will the growth hormone treatment stop when the child has finished growing?

Adults also need growth hormone, but they need far less than a child. Some young people are found to have extremely low levels of growth hormone and go back onto treatment on an adult dose. Others are found not to need this any longer because the produced GH exceeds the necessary levels for adults.

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11. Apart from growth hormone, what happens with hormone replacement in adulthood for patients with multiple deficiencies?

Hormone replacement continues regardless of growth hormone treatment. The prognosis for the patients is particularly good, if the treatment starts early and is carefully monitored by a specialized center.

12. Useful links for parents

https://www.pituitary.org.uk/

https://childgrowthfoundation.org/