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SOD. Septo-optic dysplasia

NIHR BioResource – Rare Diseases study project

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Summary

Septo-optic dysplasia is the name given to the condition where a child is diagnosed with two or more of the following problems: optic nerve hypoplasia, midline brain abnormalities and pituitary gland abnormalities. It is a rare condition affecting around 1 in every 10,000 births, with boys and girls affected equally. Septo-optic dysplasia is a congenital condition so it is present at birth, although it may not be diagnosed until childhood, or rarely, adolescence. Septo-optic dysplasia was previously known as de Morsier syndrome.

There is some evidence to suggest that septo-optic dysplasia is caused by a mutation or change affecting one particular gene (a small part of one of your chromosomes). However, in the majority of cases, it is not thought to be an inherited disorder passed on from parent to child. It is highly unlikely to recur in further children within the family, and this suggests that the cause of the condition is complex. It has been shown to occur more frequently in younger mothers, and environmental factors may play a role. There may have been a particular problem within the pregnancy, which is highly unlikely to recur in future pregnancies. More research is needed to confirm or rule out these theories.

As septo-optic dysplasia affects a variety of body systems, a multidisciplinary approach involving different specialists is required to ensure that the best treatments are given. Additionally, in addition to hormone deficiencies, sleep disorders and autism are particularly frequent. The team may include endocrinologists (hormone specialists), ophthalmologists (eye specialists) and neurologists (brain specialists). Input from experts in visual impairment and developmental paediatricians, as well as from specialists in sleep disorders and neuropsychiatry, may be required.

The basis of treatment is to identify which hormones are absent or not being produced properly, and replace these with man-made versions. Additionally, treatment may be required to help with the sleep disorders and behavioural issues.

Collaborators:

- The Developmental Vision Clinic[1] at Great Ormond Street Hospital (GOSH)
- Evelina London Children's Hospital
- Maudsley Hospital - CIPRD
- Birmingham Children's Hospital
- Royal Manchester Children's Hospital
- Moorfields Eye Hospital

We have support from the NIHR BioResource to gather a cohort of SOD patients. The main aim of this project is to gain a better understanding of the condition and why it happens. This will then help us improve the treatment for this highly complex condition which is associated with severe lifelong morbidity. To achieve these objectives, we will collect DNA samples and detailed phenotyping data from recruited patients.

Recruitment Criteria

Inclusion

Major abnormalities

- Classic optic nerve hypoplasia/eye defects such as coloboma
- Midline brain abnormalities
- Pituitary gland abnormalities
- Microphthalmia
- Anophthalmia
- Hypopituitarism

Minor abnormalities

- Congenital abnormalities of the brain
    - Cysts
    - Schizencephaly
    - Grey matter heterotopia
    - Hydrocephalus/ventriculomegaly

References

[1]https://www.gosh.nhs.uk/health-professionals/clinical-specialties/neurodisability-information-health-professionals/clinics-and-services/developmental-vision-clinic