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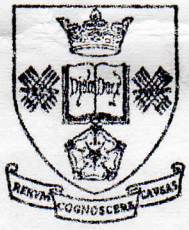

CONGENITAL ANOMALIES RESEARCH UNIT

DEPARTMENT OF CHILD HEALTH

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CONGENITAL ANOMALIES RESEARCH UNIT

The Congenital Anomalies Research Unit of the Department of Child Health has been built through the generosity of Sir Isaac Wolfson. The study of illness and disability which is congenital has become relatively more important as most of the common acquired diseases can be prevented or adequately treated.

During recent years the investigation of spina bifida and of hydrocephalus has been a major part of the research effort in the Department of Child Health and at the Children's Hospital in Sheffield, and it was the need for further intensive research into these congenital conditions that prompted the Department to look for a generous benefactor so that accommodation and equipment should be available for research workers.

The Research Unit is therefore primarily, but not exclusively, designed for the investigation of spina bifida, hydrocephalus and their related disorders. Work that has been done by the team in the past is being collated and assessed as the starting point for the extensive programme of research which this building now makes possible.

Although a particular project of one research worker must be pursued in depth, the team keeps constantly in view the plan of each investigation in the whole pattern of research into spina bifida.

Before outlining this research plan it is necessary to give a brief account of spina bifida.

The serious type of spina bifida, for which research is so necessary, is a condition in which the nervous tissue of the lower part of the spinal cord lies exposed on the surface of the baby's back at birth. The bony arch which normally covers the spinal cord is split widely open and there is not even a layer of muscle or skin to protect the nervous tissue.

Parts of this nervous tissue of the spinal cord may be abnormal at birth leading to a weakness or loss of power in some muscles of the legs, but parts of the cord may be damaged or infected after birth and early operation may prevent this further loss of activity.

The paralysis or weakness of the lower limbs is usually accompanied by some weakness or paralysis of the bladder and imperfect control of the bowels. These complications are now occupying a more important position in research to prevent the long term illnesses due to kidney disease in some of these children.

Most babies have excessive accumulation of the cerebrospinal fluid which normally lies within and around the brain, and it is the possibility of treating this complication (hydrocephalus) that has made possible the concerted plan of treatment of the whole child. The treatment of hydrocephalus still presents many unsolved problems.

SUMMARY OF RESEARCH PROGRAMME

1.	Basic Studies:	Incidence, Genetics, Epidemiology, Family Histories. Embryology Pathology
2.	Spina Bifida	Methods of timing of operation and the results of treatment.
3.	Infection:	Local Infection Meningitis Prevention and Treatment with New Drugs
4.	Hydrocephalus:	Early Diagnosis Treatment and Results Effect on Sight and Intellect
5.	Kidney & Bladder Function:	Prevention and Treatment of Infection Indications for Operation Results of Treatment
6.	Orthopaedic Problems:	Study of Causes of Deformity Development of new techniques to enable children to walk.
7.	Social, Psychological and Educational Studies.	

COMMENTS ON SOME ASPECTS OF RESEARCH

1. Basic Studies

   (a) Frequency, genetics and epidemiology.
   Detailed family studies have been done over the past 10 years: Results have already been presented but further analysis of the 10 year series will be needed.

(b) **Embryology:**
A study is in progress of the earliest stages of of the development of spina bifida in the embryo.

A further study is planned of the development of the nerve supply of the bladder and ureter.

(c) Pathology:
During the past 10 years there have been investigations into:

i) the site of maximum damage to cerebral tissue in hydrocephalus.

ii) the late effects of treatment on the volume of grey and white matter in the brain.

iii) the abnormal development of the blood vessels around the base of the brain.

iv) the variation in numbers and position of nerve cells in the spinal cord.

Studies have now been started on the detailed nerve pattern of the bladder and ureter in established cases of spina bifida.

2. <u>The Spina Bifida</u>

i) The study of early repair of spina bifida.

ii) The use of antibiotics in preventing infection of the spinal wound.

iii) Variations in the spinal lesion.

Hemimyelocele: Where only half the spinal cord is exposed on the surface.
Diastematomyelia: Where the cord is split in two by a bone spur.

3. <u>Meningitis</u>

i) Prevention.

ii) Study of new drugs in treatment.

iii) Study of the concentraion of drugs in cerebrospinal fluid.

iv) Results of treatment: although meningitis is an increased hazard, vigorous and intensive treatment may cure it with no after effects.

4. Hydrocephalus

i) It is known that severe hydrocephalus requires treatment and also that mild hydrocephalus may never cause trouble and may be better without operation. The selection of cases requiring operation presents many difficulties and is helped by follow up studies of patients treated from 1959 onwards.

ii) In addition new techniques of assessing the quantity and pressure of fluid within the skull are being developed. These include ultrasonic examination and also the production of a minute pressure recording device to detect pressure variations within the brain.

iii) Late results in relation to intellectual development.

iv) The use of drugs to reduce the pressure of fluid within the brain is also being reviewed.

v) Severe hydrocephalus may produce blindness. Research is being carried out to determine the chances of complete recovery of sight if treatment is undertaken.

5. Kidney and bladder function

The basic problem is the failure of normal emptying of the bladder; this may lead to stagnant urine and infection, or it may cause back pressure on the kidneys and damage to the kidney tissue.

Infection causes chronic ill health but may also contribute to permanent kidney damage: several intensive trials of various drugs to control this infection have been undertaken and are being continued.

Intensive studies are directed towards detecting early signs of poor function by X-ray studies, by isotope studies and by measuring the pressure within the bladder.

Early diagnosis will lead to early treatment but the best type of treatment is still not clear and the long

term results of various operations to improve bladder emptying are being studied.

Sometimes the bladder can no longer be used and substitute drainage of the kidneys provided (ileal loop). The long term study of these substitutes is in progress.

The possibility of stimulating a paralysed bladder by drugs or electronic implants is being actively pursued.

6. Orthopaedic Problems

Weakness or paralysis of some muscles may cause dislocation (for example of the hip) and a close investigation of the early cases showed that if the active muscles were transplanted the hip could be replaced and kept in position and the child enabled to walk. This research has led to a profound improvement in outlook for spina bifida children.

Muscles which appear to be paralysed may have the potential for recovery: special electronic study of these muscles (electromyography) is being undertaken.

Many have deformed feet and although the position can be corrected, it is still not clear whether this is best done when the infant is very small or when about to get onto his feet.

The results of the two methods of treatment have been the subject of a detailed review.

Some babies, even at birth, have such a severe spinal deformity that at the time of the primary operation immediate correction of the bony deformity is attempted.

7. Social, psychological and educational studies

The education of spina bifida children is of great importance; they are more dependent on education for their future role in the community than are normal children.

An educational psychologist is studying the pattern of intellectual development in children of 4 years and older, and there is some evidence that they have a relatively greater facility with speech than other children.

Close contact with educational authorities is maintained to find the optimal type of education for each child.

This research has been supported from various sources:

The Endowment Fund of the United Sheffield Hospitals

The Richard Fund for Children's Research (including donations from local organisations)

National Fund for Research into Crippling Diseases

The Pharmaceutical Industry

University Grants Committee

Private Benefactors

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