

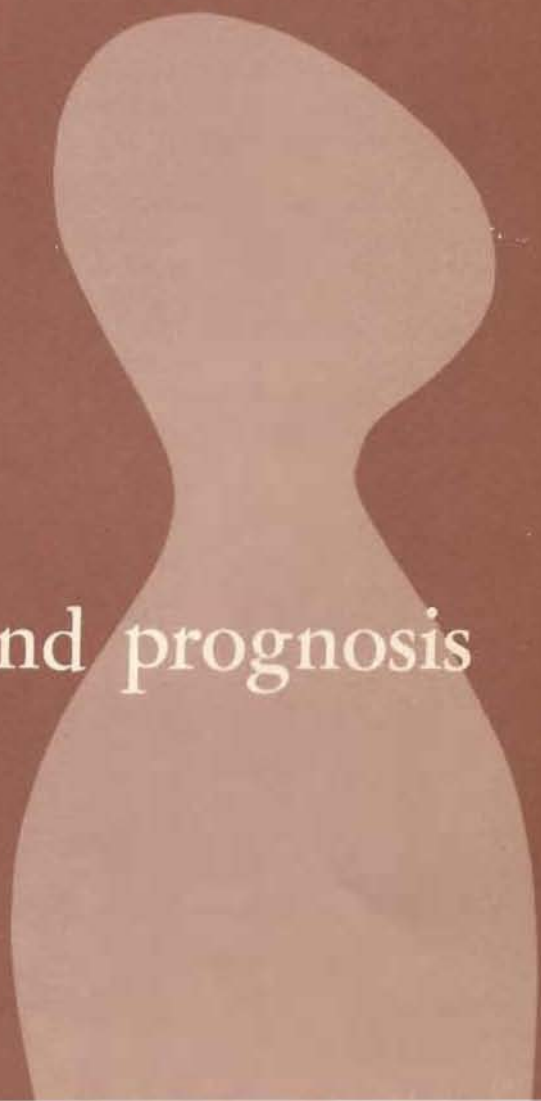
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the  
care  
of  
the

RETARDED  
CHILD

therapy and prognosis



## FOREWORD

Often mental retardation is not the only handicap a retarded child suffers. Some of these children have difficulty in walking, with vision, in hearing, or with other neurological disorders.

The material presented at the Seventh Annual Arthur Parmalee, Sr. Child Development Institute held in April 1962 at the Childrens Hospital, Los Angeles, California, constituted a collection of valuable information that could help in understanding some of these children—and it is for this reason that the Children's Bureau wished to make it available to a wide audience.

The purpose of the Institute was to provide training to the professional persons in attendance. Subsequently, the material was used by the Childrens Hospital in the training of students. The sponsor of the Institute, the Child Development Clinic of the Childrens Hospital, is supported by a grant from the Children's Bureau, through the California State Health Department.

Dr. Arthur Parmalee, Sr. began this series of institutes in 1955 to inform physicians about the advances being made in the field of mental retardation.

He was intensely interested in mentally retarded children and played an active role in establishing the present Child Development Clinic at the Childrens Hospital of Los Angeles.

At the time of his death in June 1961, Dr. Parmalee was still actively teaching students, participating in clinic demonstrations for the resident physicians at Childrens Hospital, writing articles, and seeing patients in his office.

The Children's Bureau joins the staff of the Child Development Clinic in hoping that some of his spirit has been captured in this report.

Katherine B. Ottinger

Katherine B. Ottinger  
Chief, Children's Bureau  
WELFARE ADMINISTRATION

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Proceedings of the  
Seventh Arthur Parmalee, Sr.  
Child Development Institute

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U.S. DEPARTMENT OF HEALTH, EDUCATION, AND WELFARE  
WELFARE ADMINISTRATION • Children's Bureau • 1964

## CONTENTS

### INTRODUCTION TO THE PROBLEM OF MENTAL HANDICAPS: NORMAL GROWTH PATTERNS

Ada Morton, R.N. .... 1

### DIAGNOSTIC AND THERAPEUTIC PROCEDURES FOR THE TREATMENT OF NEUROSURGICAL PROBLEMS IN CHILDREN

Frank Anderson, M.D. .... 7

### THE ROLE OF THE NEUROLOGIST IN THE DIAGNOSIS OF RETARDATION

Robert P. Sedgwick, M.D. .... 12

### OPHTHALMOLOGIC PROBLEMS

Warren A. Wilson, M.D. .... 23

### NEW DEVELOPMENTS IN GALACTOSEMIA

George N. Donnell, M.D. .... 31

### THE PREDICTABILITY OF GESELL DEVELOPMENTAL SCALES IN DOWN'S SYNDROME

Jack Share, M.A.; Betty Galiker, M.S.W.; and  
Richard Koch, M.D. .... 39



# INTRODUCTION TO THE PROBLEM OF MENTAL HANDICAPS:

## Normal Growth Patterns

ADA MORTON, R.N.

Librarian, California Council for Retarded Children

WHEN A CHILD IS BORN, some parts of his body are more developed than others. Normally, the skull of the newborn is large in proportion to the rest of the body. Figure 1 illustrates this by comparing the relatively large abdomen, the long trunk, and the short arms and legs of the infant (1).

By the time the baby is a year old, his body from neck to feet is approximately  $3\frac{1}{2}$  times the length of his own head. His legs are still quite short compared to the length of his head and torso. This makes the center of his body at a line through the abdomen rather than at the crotch. The 1-year-old can be measured in terms of his own head. Using this measurement, he is about  $4\frac{1}{2}$  heads high (figure 2).

As the child grows older, his legs and arms become much longer. By the time he is 8, the center of his body has moved down to a line above the hips. Again, the head can be used as a unit of measure. The head has grown somewhat larger and using the new head size, the 8-year-old is about  $6\frac{1}{4}$  heads high. At the age of 12, he is about seven heads high, with the center of his body at the crotch. From this period on, all the parts of the body will develop at about the same rate, until the figure reaches its full development at approximately 25 years. Some children tend to grow much more rapidly than others, but the proportions given here are a fairly accurate guideline of skeletal development.

### *Cranial sutures*

The vault of the skull is made up of four flat bones which are connected by soft membranes.

Because these bones are not united, they can be molded to adapt to the mother's pelvis during labor and thus pass through it with little resistance and injury. After a prolonged labor, the head is sometimes drawn out to a rather long shape. Within a few weeks after birth the head tends to resume a more rounded, symmetrical form. At birth the skull measures approximately 13 to 14 inches in circumference. It grows rapidly during the first year to attain

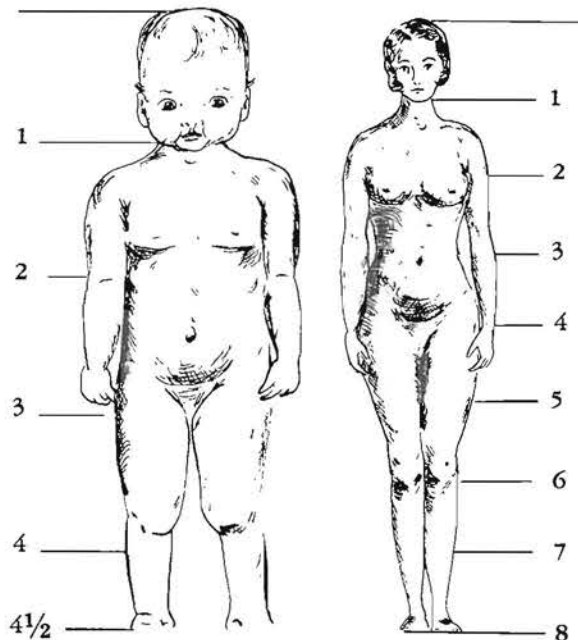


Figure 1.—ANATOMICAL COMPARISON OF INFANT AND ADULT USING THE HEAD MEASUREMENT IN DRAWING TO IDENTICAL SCALE.

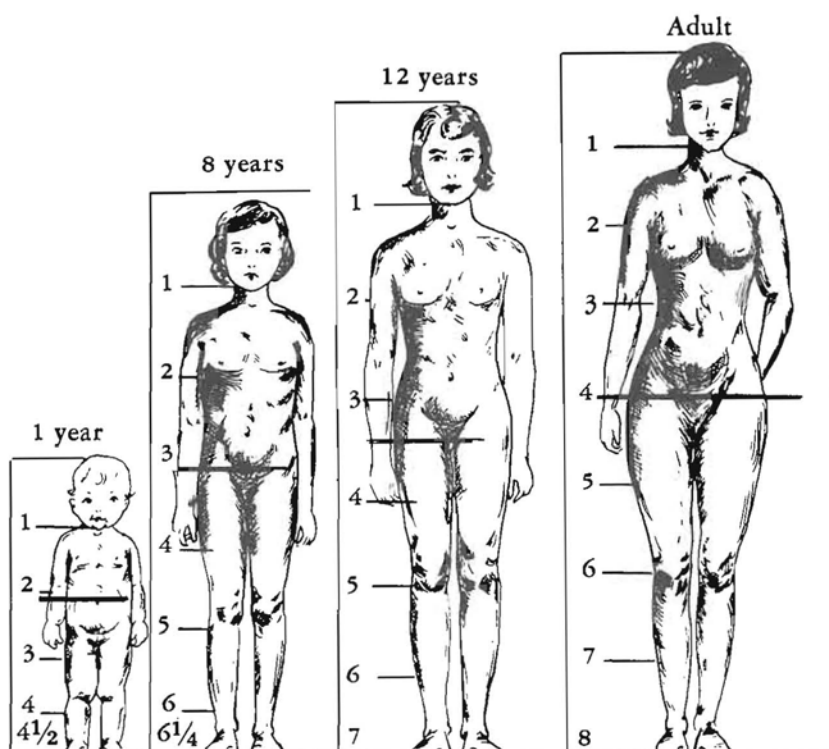


Figure 2.—NOTE DROP IN MIDLINE AS SKELETAL GROWTH MATURES.

a circumference of approximately 18 inches. By the age of 5 years the child's head circumference measures 20 to 21 inches and grows very little thereafter. The brain has attained most

of its physical growth by the age of 3 or 4 years. (2).

Figure 3 identifies the cranial sutures as seen from the side. The lambdoidal suture (1)

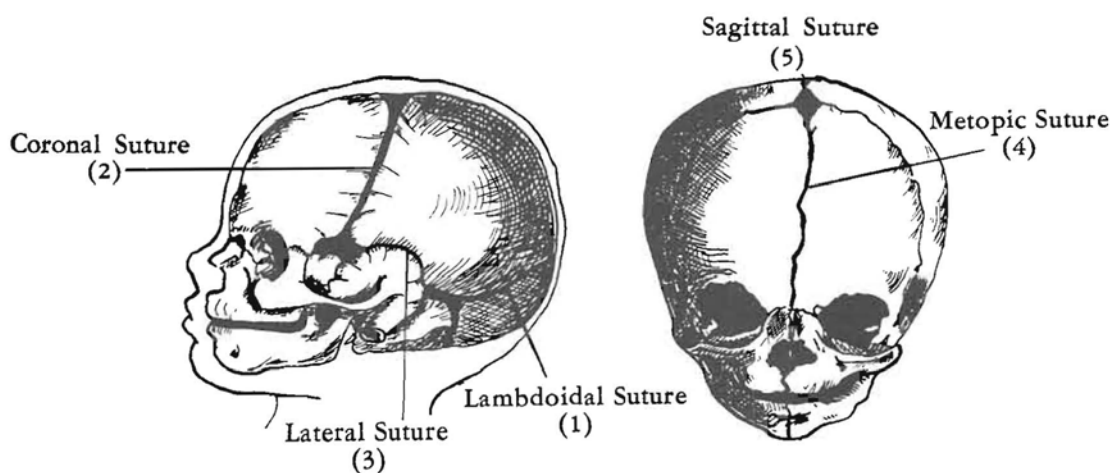


Figure 3.—CRANIAL SUTURES IN THE NEWBORN.

is located between the parietal and occipital bones. The coronal suture (2) is located between the frontal and parietal bones. The temporal or lateral suture (3) is located between the temporal and parietal bones. The metopic suture (4) lies in a midline position between the two frontal bones. The sagittal suture (5) is also in midline, and separates the two parietal bones.

## Fontanels

At the point where the two frontal and two parietal bones meet lies an open, diamond-shaped space filled in by a membrane called the anterior or "large" fontanel. Normally this fontanel closes between the ages of 10 and 18 months. The posterior or "small" fontanel is located at the juncture of the parietal and occipital bones. This is not an actual opening; it is the meeting of the three sutures identified as sagittal, coronal, and metopic. Normally this fontanel closes between the second and third months after birth.

Figure 4 shows the top of the skull and indicates the posterior or "small" fontanel; Y-shaped at upper pole (1); the anterior or "large" fontanel, diamond-shaped at the sagittal suture (2) in the midline (3); the coronal sutures which proceed laterally from the large

fontanel (4); and the metopic or frontal suture leading down from the large fontanel (5).

## Normal growth pattern of the skull

At 1 year, the head is still large in proportion to the face and features which seem enclosed by chubby, full cheeks. The eyes are placed low on the head. The chin and nose are quite small, and the neck is short and fatty.

At 6 years the head is still smaller in proportion to the face which is growing larger with the chin becoming more pronounced. The mouth and the nose show a more definite shape and the neck is growing longer.

At 11 years a pronounced change has taken place. The head is definitely smaller in proportion; the face has lengthened with the jaw and chin becoming quite definite. The nose has grown longer and the eyes are placed higher. The mouth assumes a firmer quality and the neck begins to develop.

At 17 adult proportions are becoming evident. The eyes, lips, nose, and chin have developed almost to their full size. The jaw and cheekbones assume much more prominence and unique facial characteristics begin to form.

In the adult, the cranial bones are interlocked together like teeth. There is no longer

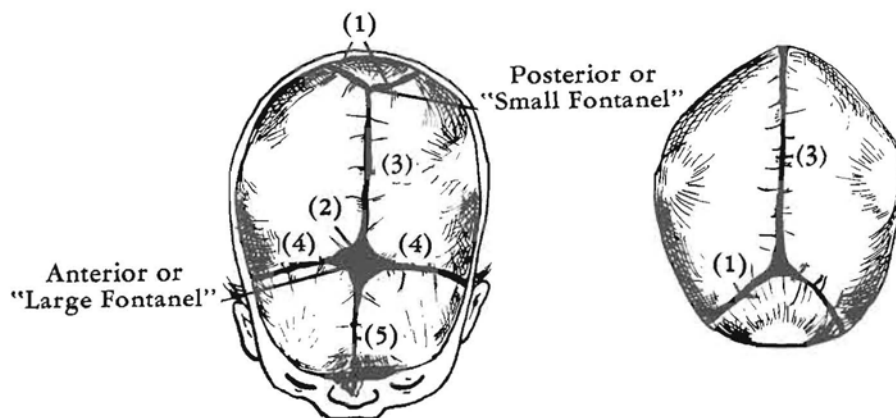


Figure 4.—FONTANELS IN THE NEWBORN.

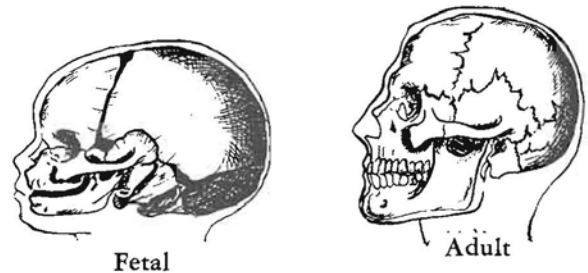
any stretch or any significant play between the bones. Figure 5 compares the fetal skull and the adult skull.

### *The ventricular system and the cerebrospinal fluid*

The *ventricles* are cavities in the brain (figures 6 and 7). There is a lateral ventricle in each half of the brain under the mass of white fibers which connect the two hemispheres or halves of the brain. These connecting fibers are called the *corpus callosum*. The third ventricle is behind the lateral ventricles but is connected with each one by means of small openings called *foramina of Monro* (so called because they were first identified by Scottish anatomist Alexander Monro, 1697–1767). The fourth ventricle is in front of the cerebellum, behind the pons varoli, and the medulla. The third ventricle communicates with the fourth by means of a slender canal called the *aqueduct of Sylvius* (named for the French anatomist Jacobus Sylvius, 1478–1555). In the roof of the fourth ventricle are three openings, the median one is called the *foramina of Magendie* (Francois Magendie, French physiologist, 1783–1855). The other two are called the *foramina of Luschka* (Herbert Luschka, German anatomist, 1820–75). By means of these three openings the ventricles communicate with the subarachnoid space, which is located between the arachnoid mater and the pia mater which in turn are composed of a spongy connective tissue. This tissue contains communicating channels in which the subarachnoid fluid circulates (3).

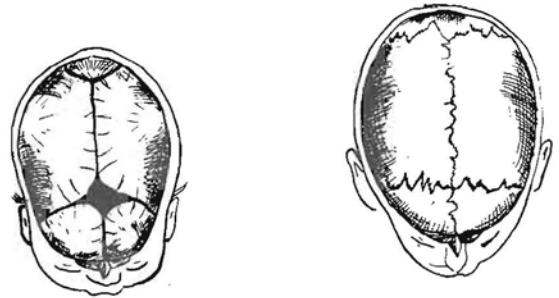
The brain and spinal cord are enclosed within three membranes which, named from the outside, are *dura mater*, *arachnoid mater*, and *pia mater*. The *dura* is the tough outer layer which is adherent to the bones of the skull. The inner (meningeal) layer of the *dura* covers the brain and sends numerous folds inward for the support and protection of the different lobes. The *arachnoid mater* is a delicate membrane placed between the *dura* and *pia*. The cranial portion of the *pia mater* covers the surface of the brain and dips down between the convolutions (3,4).

The *choroid plexus* (figure 6) is located



Illustrating spaces between cranial bones of skull.

Illustrating toothed or dentated sutures of rigid skull.



Illustrates how cranial bones can overlap permitting passage through the birth canal.

Adult skull viewed from above illustrating the interlocking suture lines.

Figure 5.—COMPARISON OF SUTURES BETWEEN THE FREELY MOVABLE NEW-BORN SKULL AND THE RIGID ADULT SKULL.

within the ventricles and produces the cerebrospinal fluid; this mechanism is as yet not entirely clear.

### *Cerebrospinal fluid*

The ventricles of the brain, the central canal of the spinal cord, and the spaces of the arachnoid between the pia and dura mater are all filled with a clear colorless fluid. This fluid is secreted into the ventricles of the brain by the choroid plexus. After filling the lateral ventricles, the fluid escapes by the foramina of Monro into the third ventricle and then by an aqueduct into the fourth ventricle. From the fourth ventricle, the fluid pours through the

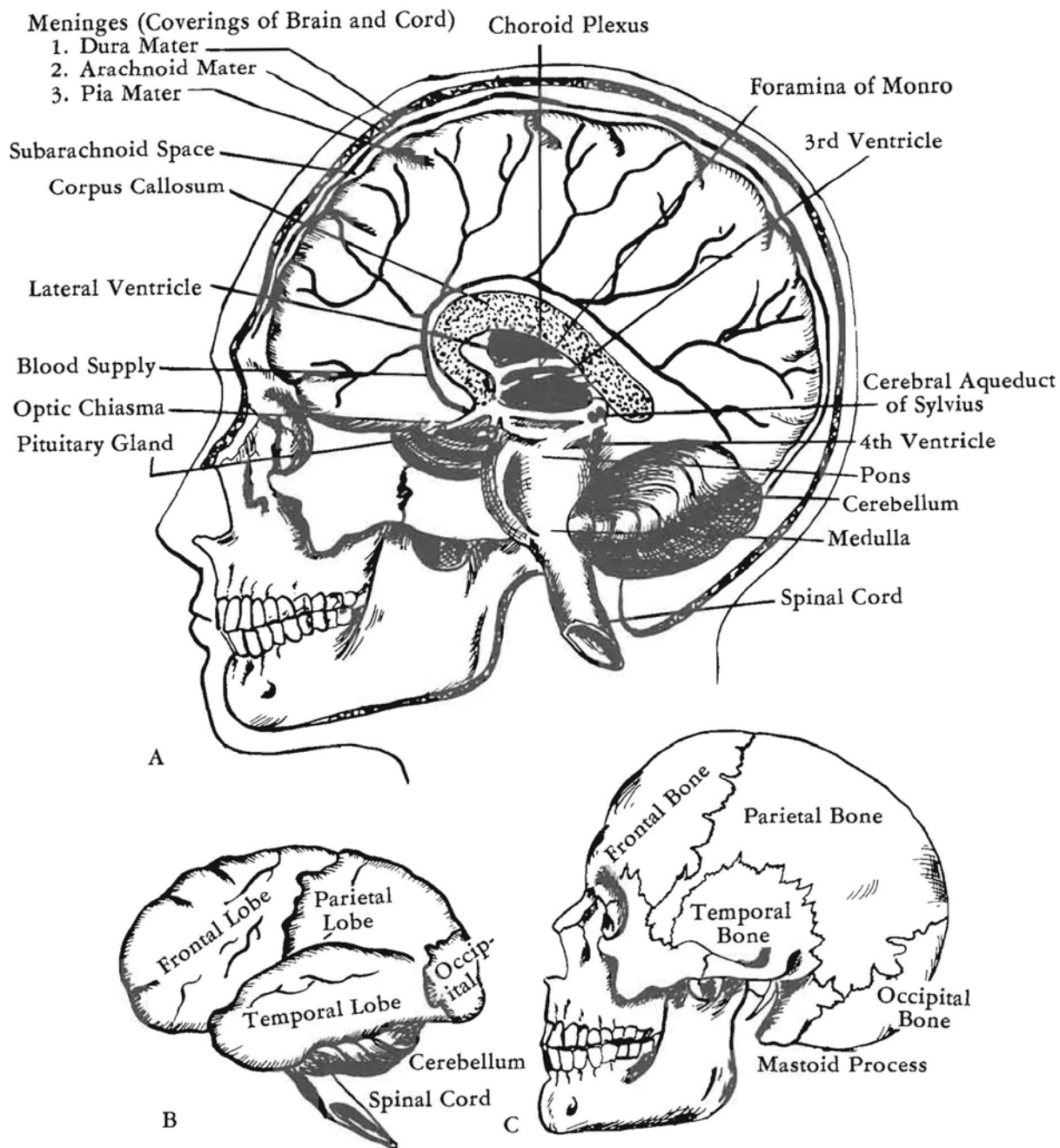


Figure 6.—A. BRAIN AS SEEN IN SAGITTAL SECTION OF HEAD. B INDICATES THE LOBES IN RELATIONSHIP TO THE CRANIAL BONES IN C.

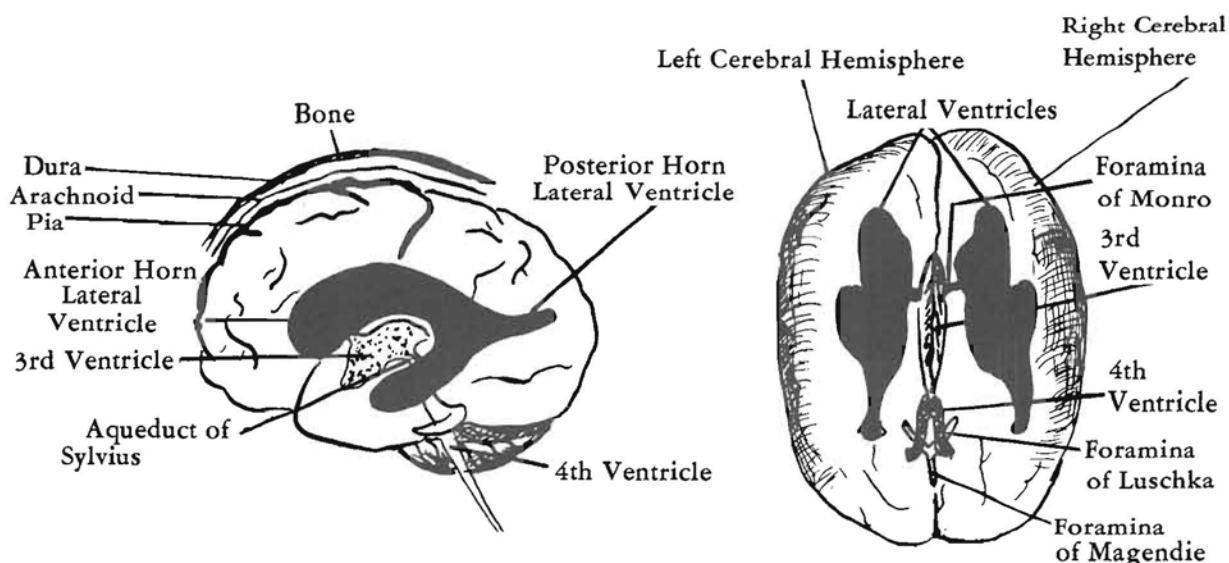


Figure 7.—“TRANSPARENCY” TO SHOW OUTLINES OF CEREBRAL VENTRICLES.



Figure 8.—ILLUSTRATING THE PATHWAY OF THE CEREBROSPINAL FLUID FROM VENTRICLES.

“Birth Defects,” Morris Fishbein, M.D.; Lippincott Co., 1963.

foramina of Magendie and Luschka into the subarachnoid space. From here the fluid circulates inferiorly in the spinal canal within the arachnoid space and returns upward in the subarachnoid space. It bathes all portions of the brain and in this way serves the purpose of keeping the brain and spinal cord moist, lubricated, and protected from varying pressures (see figure 8).

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# DIAGNOSTIC AND THERAPEUTIC PROCEDURES FOR THE TREATMENT OF NEUROSURGICAL PROBLEMS IN CHILDREN

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THE MAJOR TYPES of neurosurgical lesions known to have a direct bearing on the development and function of the child are: (1) hydrocephalus—water on the brain, (2) crani-  
osynostosis—premature closure of the cranial sutures, (3) subdural hematoma, (4) brain abscess, (5) brain tumor, (6) vascular anomalies, and (7) seizure foci.

The most encouraging factor about neurosurgical lesions is that there is a reasonable chance for improvement through surgery or some specific kind of treatment method. But this is possible only if the disorder is not too severe or is not unduly advanced. Unfortunately, there are many babies with such problems that the medical profession cannot help because their condition is highly developed by the time they are born and therefore is not remedial.

For the ventricles to be visible on the X-rays, they must contain air as a contrast media. The procedures for introducing air into the ventricles are: (1) pneumoencephalogram (pneumo—gas, encephalogram—brain picture) and (2) ventriculogram. In these procedures some of the cerebrospinal fluid is removed and replaced with sterile air. In a pneumoencephalogram the patient is placed in an upright position and a spinal puncture performed. Since air is lighter than cerebrospinal fluid, it rises and accumulates in various portions of the brain. The ventriculogram is performed by inserting a sterile needle directly through the cortex of the brain into the ventricle. Air is then introduced and subsequent X-rays taken. Thus, diagnosis of certain malformations and

diseases of the brain can be made. Children are usually prepared for this operation by receiving an anesthetic and will sleep for a period of time afterwards (1).

## *Hydrocephalus*

A large head is not always due to excess fluid in the brain. The head may be large for a variety of reasons. In fact, some people simply have unusually large heads as a familial trait.

Hydrocephalus in infancy and childhood is due to an abnormal increase in the volume of cerebrospinal fluid within the skull. This fluid is formed and absorbed so that in normal circumstances the volume remains constant while circulating freely throughout the system. A distinction is made between increased volume with and without increased pressure. When hydrocephalus occurs *without increased* pressure it is merely compensatory to atrophy (shrinking) or a maldevelopment of the brain which is termed hypoplasia. This type of hydrocephalus is "external hydrocephalus." When hydrocephalus is accompanied by an *increase* in pressure, it is indicative of a disturbance in the absorption, formation, or circulation of the cerebrospinal fluid occurring either singly or in combination and is termed "internal hydrocephalus." Both these difficulties may be due to congenital cerebral abnormalities or they may occur after birth as a result of trauma, infection, or abnormal growths.

The circulatory system for the cerebrospinal fluid can be divided into two parts: (1) ventricular system and (2) subarachnoid spaces. The ventricular system is made up of the four ventricles discussed earlier. The subarachnoid spaces radiate out like a sunburst over the surface of the brain. These spaces originate at the base of the brain and carry the cerebrospinal fluid from the openings of the fourth ventricle forward and upward over the cerebral hemispheres (see figure 8).

Obstruction in the cerebrospinal fluid pathway is a much more frequent cause of hydrocephalus than is failure in the absorption mechanism. A common cause of obstruction is occlusion of the aqueduct of Sylvius by a congenital maldevelopment or by a post-infective complication (2). Obstructions in the foramina of Monro, Magendie or Luschka are usually due to an acute or chronic adhesive meningitis, but they also may be due to a malformation such as a septum extending over the inferior part of the roof of the fourth ventricle (2).

Failure of adequate absorption can be due to an impediment in the upward movement of the cerebrospinal fluid such as adhesions from meningitis obstructing the normal passageways for the circulation of the fluid. Deformity of the bones at the base of the skull has also been suggested as a possible cause of the condition.

Hemorrhage at birth has been known to cause hydrocephalus through blockage of the aqueduct of Sylvius or the subarachnoid space (2). The occlusion in such cases may be immediate or may follow later with the formation of adhesions in the area of the clot.

There are some conditions in which the outward appearance of the enlarged head appears to be due to hydrocephalus but is not. The enlargement may be due to blood clots (chronic subdural hematoma) over one or both hemispheres. In some instances the volume of old blood may be five times the size of the brain. Tumors and abscesses also cause increased pressure which enlarges the head.

Another cause of hydrocephalus is a deformity of the spine called meningocele (figure 9). This is a hernial protrusion of the meninges or membranes covering the cord (3). In this condition the spinal nerves are out-pouched

from the spine and enclosed in a sac. This may occur at any point along the spine but usually at the neck or lower back. Hydrocephalus accompanies it in a certain proportion of afflicted infants. Involvement in the condition may

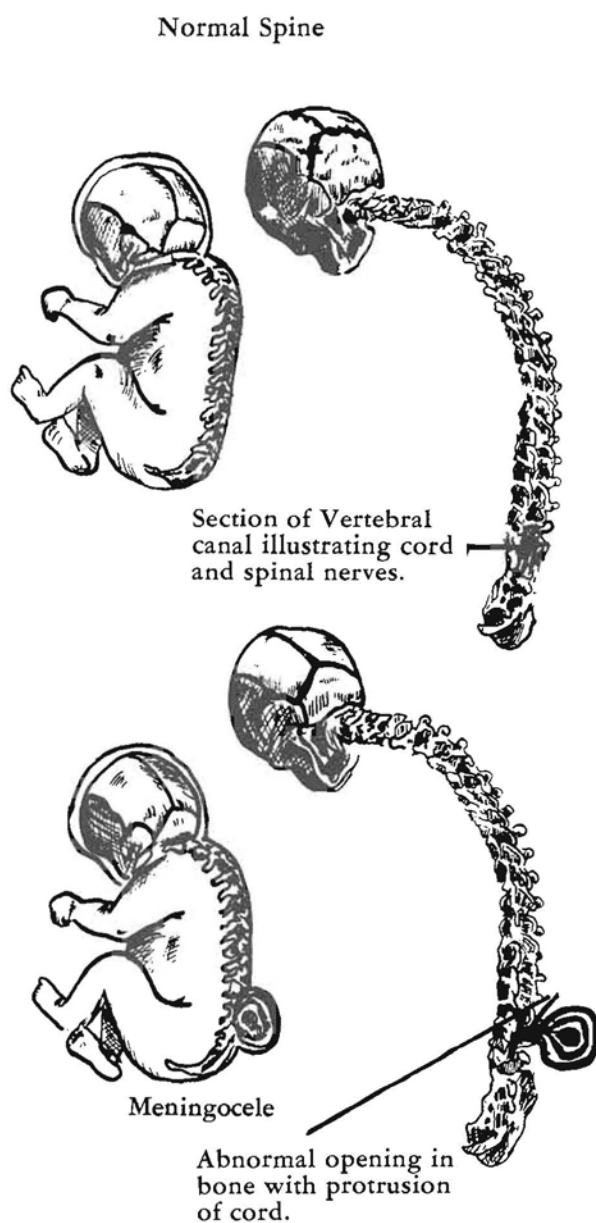


Figure 9.—ILLUSTRATING THE NORMAL AND ABNORMAL DEVELOPMENT OF THE SPINE.

include paralysis of the lower extremities, poor bowel and urine function, and retarded mental and physical development.

### *Clinical picture of hydrocephalus*

Early diagnosis is difficult. Repeated observation of the rate of head growth may give the doctor early indication of abnormality (figure 10). As fluid accumulates in the ventricles, the brain enlarges due to increased tension. The cortex of the brain may become paper thin due to pressure causing the skull to turn to enlarge. As stated earlier, the sutures gap widely, the fontanels enlarge and the circumference of the skull increases. The skull tends to become globular with an accompanying alteration in the shape of the face so that the eyes become wide apart and the bridge of the nose depressed. Some degree of optic atrophy and visual impairment may develop. Deafness is an occasional complication seen in hydrocephalus due to postmeningitis cases (2).

Hydrocephalics may have seizures and spasms which usually affect the lower limbs more than the upper. The severity of the physical or mental involvement is dependent on the extent of cranial displacement and involvement (2). Some cases require 24-hour care due to severe disability while others are able to participate in normal daily activity, including school.

If the onset of hydrocephalus is delayed until after closure of the sutures there is little likelihood of enlargement of the skull as seen in figure 10. Diagnosis must be made on the basis of the patient's history, physical condition, and pneumoencephalograms.

Generally a distinction is drawn between progressive and arrested hydrocephalus. In *progressive or expanding* hydrocephalus there is progressive mental and physical deterioration usually terminating in death. In *arrested or stationary* hydrocephalus, once the expansion has stopped, the intracranial pressure stabilizes and some degree of improvement may follow. The sutures fill in and the skull may become rigid and thick. It is not clearly understood what happens at this point. There appears to be a period of balance between the secretion of

the spinal fluid and its absorption (2). Mental capacity may be unimpaired in these cases.

### *Treatment*

The surgical treatment of hydrocephalus consists of reducing the amount of cerebrospinal fluid produced or the rerouting of the fluid around the point of obstruction.

For many years the treatment of hydrocephalus was to surgically open the skull and remove the choroid plexus located in the lateral ventricles. This is the main site for the manufacture of cerebrospinal fluid.

The present treatment of this condition when there is no tumor or other specific cause is the *Shunt method*. This is a rerouting of the fluid from one part of the cerebrospinal fluid pathway to another tissue where the fluid can be reabsorbed (4).

Figure 10 demonstrates one Shunt method. Here excess fluid from the ventricle is conveyed into the blood stream by means of a small tube, approximately one-eighth of an inch in diameter. At the end of the tube is a minute valve, approximately 2 millimeters in diameter, with a small slit in it allowing the spinal fluid to go out but preventing the blood from entering. This method was designed by Mr. W. Holter, a Philadelphia engineer and parent of a hydrocephalic boy. An air study would show the tube inserted into a ventricle of the brain, brought out through a hole in the skull, then running down under the scalp and down into the heart. Other procedures have been used, but are largely superseded by this newer technique.

### *Craniosynostosis*

In rare instances premature synostosis (the union of bones by means of osseous substances) of the cranial sutures occurs as a congenital trait which results in deformity of the skull. In craniosynostosis, when there is compression of the brain, X-rays reveal convolutional atrophy of the brain, absence of one or more sutures, shallow orbits and underdevelopment of the sinuses, in addition to the abnormal shape

Enlargement due to accumulation of fluid within the ventricles or cavities.



A



B

Surgical treatment draining excess fluid via Shunt method



C

Figure 10.—A AND B ILLUSTRATE THE CLINICAL PICTURE OF HYDROCEPHALUS. C SHOWS ONE OF THE VARIOUS SHUNT METHODS USED TO DRAIN EXCESS ACCUMULATION OF CEREBROSPINAL FLUID FROM VENTRICLES.

of the skull. The most common abnormal fusion is at the sagittal suture joining the two parietal bones across the vertex of the head. According to data gathered on 102 cases seen at the Childrens Hospital of Los Angeles, between 1954 and 1962, this type of fusion was seen in approximately 63 percent of the cases. This suture ordinarily is open to let the brain grow for at least 12 years. In some babies however, it is fused at birth. Thus the head is unable to broaden in growth, so it becomes elongated and appears pointed. The condition is termed *scaphocephaly*, or "boathead." This elongation is due to the growth of the forehead and the back of the head.

When the coronal or crown suture fuses prematurely, the head cannot elongate. This condition, *brachycephaly*, gives the head the appearance of being very short, broad and high. In the event that one side of the coronal suture

fuses, that side of the forehead will not grow. The bone is flat and much smaller and as a result there is elevation of the eyebrow and some recession of the eye on the affected side.

*Trigonocephaly* results from the fusion of the metopic suture between the two frontal bones producing a head that is triangular in shape. The skull maintains the appearance of a fetal or prenatal skull.

*Oxycephaly* occurs when all the sutures fuse prematurely. The joints of the skull are fused solidly and the brain does not grow adequately, resulting in mental and physical retardation.

### *Surgical treatment for craniosynostosis*

If surgical repair is done before there is significant cerebral or visual damage from compression of the brain, defects of vision and intel-

lect can often be avoided. Various techniques have been developed to improve the surgical channels and to keep them open longer. In essence, this is a means of creating room for the growing brain by cutting false joints along the prematurely closed suture. This provides a joint so that the head can elongate or permit the head to grow transversely. After the channels have been cut, an agent that slows the regrowth of the bone (polyethylene film) is applied to the cut surface of the cranial bones. This operation works quite well if it is done in time. It permits the skull to expand normally and prevents retardation. As a general rule, surgery is performed not later than 3 months after birth although each case varies with the kind and severity of the problem. If there are coronal or multiple synostoses, surgery should not be delayed. The decision is more difficult in cases of isolated sagittal synostosis, since some children with this anomaly remain symptom-free. Early is the key word. Some

varieties of skull deformities are often accompanied by mental defect complicating the outcome or prognosis for recovery with or without surgery, but those who have no basic organic difficulty recover without incident.

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# THE ROLE OF THE NEUROLOGIST IN THE DIAGNOSIS OF RETARDATION

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NEUROLOGISTS throughout the country are becoming more interested in the problems of handicapped children, especially in the areas of behavior, learning, and mental retardation. This is evident in the fact that the candidates for a board examination in neurology now have to pass examinations in Children's Neurology. In addition there is now a section on Children's Neurology in the American Academy of Neurology.

The neurologist can help in diagnosis and treatment of children with central nervous system (CNS) disorders and participate in continuing surveillance of such children with teachers, psychologists and social workers. In his initial examination, the neurologist must decide whether a child exhibits evidence of organic brain damage. If so, what is the nature of the lesion, and, if possible, what areas of the brain are involved? To do this, he must take a careful history including information on heredity, course of the pregnancy (especially illnesses of the mother during the first trimester), whether there was any bleeding during pregnancy, complications of labor, and the condition of the child at birth. Since it is estimated that some 70 percent of brain damage in children is due to genetic, prenatal, or neonatal pathology, no effort should be spared to secure accurate and detailed birth records. The neurologist notes the developmental history of the child during infancy. Was he active? What medication, if any, did he receive? Was he a feeding problem? Was there a sign of twitching or convulsions or any illness such as one of the infectious diseases of childhood—measles, chickenpox, meningitis or encephalitis? Did he ever sustain a physical

injury to the head? Did he have untoward reactions to his vaccination or immunizations? Questions regarding the child's behavior pattern are extremely important. It is noted whether a child is hyperactive, distractible, emotionally labile or has difficulty going to sleep.

After a careful history, the neurologist proceeds to a physical examination of the patient. If the patient is an infant, the early diagnostic evaluation depends greatly upon the presence or absence of the reflexes which are ordinarily manifested by all normal babies at birth (4).

**Blink reflex**—tested in each eye separately by shining a strong light directly into the eye.

**Movements of the face**—an evaluation of the integrity of the innervation of the facial musculature. If there is asymmetry of the facial structure, under both resting and active states, special attention is needed to evaluate facial movements.

**Motor activity**—an evaluation of the child's spontaneous movements as well as the activity evoked by nonspecific stimuli such as handling as to whether they are jerky or myoclonic, tremulous or jittery, writhing, asymmetrical, or convulsive.

**Extremity movements**—an evaluation of the functional range of motion in the joints of the extremities and spine as determined by observation of both active and passive movements.



**Cry**—an evaluation of the quality of the child's cry as to whether it is high pitched, stridulous, incessant, etc.

**Palmar grasp**—flexion of the fingers or normal grasp reflex elicited by touching or stroking the ulnar side (side of the little finger) of the palm of the child's hand. This reflex begins to disappear by the fourth month when more advanced hand skills are acquired.

**Plantar grasp**—flexion of the toes elicited by touching or stroking the sole of the child's feet. This appears in the fourth month after birth in some children and in almost all children by 18 months. If it persists beyond 24 months it is suggestive of pathology. Preceding the plantar grasp is the Babinski Reflex, which is the upward extension of the big toes with a simultaneous downward movement of the other toes when the sole of the foot is stroked.

**Patellar jerk**—elicited by tapping the patellar tendon or kneecap with a standard rubber reflex hammer.

**Tendo-achillis**—elicited by striking the achillis tendon with the hammer causing plantar flexion of the foot.

**Moro reflex**—a sudden outstretching of the arms as if to grasp for support elicited by a sudden loud noise or feeling of insecurity. This reaction disappears in normal infants between 3 and 5 months. Its absence in young infants may indicate injury of the brain and its persistence beyond 5 months may be indicative of brain damage.

These are the unconditioned reflexes and are the inherent, instinctive, and involuntary movements which an individual makes automatically following certain stimuli. They are present in the newborn. Even in the first few days of life muscular contractions occur when certain tendons are tapped. In the newborn there is reflex closure of the eyelid when the cornea is touched and by 1 month of age, when the eye is threatened by bringing objects close to it, the lids will automatically close. Also

present at birth is the sucking reflex when anything touches the lips. Deviations from the norms in these inherent reflexes become important diagnostic signs of delayed development.

The development of speech is further evidence of proper mental and intellectual development (1). By 5 or 6 weeks of age vocal sounds of a "cooing" nature begin. By 1 year of age, attempts to say "Mama" and "Papa" are recognizable. At 2 years of age the child has a vocabulary of approximately 300 words and is forming short sentences. The normal child learns approximately 200 to 300 words per year, but the age at which intelligible speech is acquired depends to a considerable extent upon the type of training and home environment. Absence of speech or marked retardation in talking may indicate failure of mental development, or the presence of emotional disturbances, deficient hearing, anatomic defects of the mouth, nose or larynx or some primary speech defect involving the brain but not necessarily accompanied by mental retardation.

The sensory modalities such as vision, hearing, taste, smell, the ability to differentiate between heat and cold, and to recognize the size and shape of objects are tested in as much detail as the child's cooperation permits. The cranial nerves are reviewed, with particular emphasis on movements of the palate, tongue, lips, and face, and visual field deficiencies (1).

The laboratory studies performed include skull X-rays to rule out abnormal calcifications, certain vascular abnormalities, or tumors. The structure and growth of bones are influenced by nutritional status, infection, and heredity. Under normal conditions, the stage of development of the bones, as seen in roentgenograms, offers an index of the maturity of the child (2). Other factors that might be indicated by skull X-rays are: (1) too small a skull indicating perhaps a lack of development of the whole brain; (2) an asymmetrical skull indicating an injury to the brain on one side only; or (3) an overly large head indicating hydrocephalus (water on the brain); or (4) spreading of the sutures indicating increased intracranial pressure.

The neurologist may feel that an electroencephalogram is indicated. His first job, when

confronted with a seizure, is to try to decide whether this seizure is primary (that is due to a stationary lesion), or whether the seizure is a symptom of some progressive disorder that takes precedence over the seizures. For example, if the child has a brain abscess and is having seizures as a result of it, the medical problem is not really the seizure—it is the abscess. On the other hand, if there is no such progressive treatable lesion present, then the emphasis of medical effort shifts to the actual control of the seizure. An abnormal electroencephalogram is not necessarily indicative of organic brain pathology, nor does a normal tracing rule it out (1).

Other tests the neurologist may employ would be analyses of blood chemistry, and urinalyses for abnormal chemical components such as phenylpyruvic acid, etc.

At the completion of this extensive evaluation, the neurologist's task is to try to determine whether there is any evidence of organic brain damage. If there is, what is its nature, location, extent, and prognosis? By noting and recording the child's overall behavior and maturation, he may suspect brain damage not elicited by the formal neurological examination. He may wish to send the child to other professionals such as a psychologist for further study and evaluation.

### *The electroencephalograph and seizures*

The electroencephalograph is the machine which records the electrical activity of the functioning cells of the brain. It was in London in 1929 that the possibilities of the EEG were first discussed at the old Central Pathological Laboratory in Maudsley Hospital (1). It was known that during the processes of thinking, seeing, feeling, and other activities which go on during the usual process of living, the brain gives off very minute electrical currents. Electrical impulses originating in the brain are carried by nerves to activate the whole body.

That the brain produced electric currents was discovered in 1875, by Dr. R. Caton, an English physician, while he was doing research on animals (1). In 1929, a German psychia-

trist, Hans Berger, University of Jena, demonstrated that these electrical pulsations could be "picked up" from the human scalp, amplified 1 to 2 million times and made to write a line on moving paper (1). The instrument used was called a galvanometer.

In 1934 or 1935, Edgar Douglas Adrian of the University of Cambridge gave a convincing demonstration of this method before a large audience. The apparatus has been perfected over the years, and the operation of today's elaborate apparatus has become a specialized science.

The elaborate scientific instrument of today records millions of cells beating or discharging in synchrony. What makes these millions of cells act together, or in fact, what causes a single cell to discharge, is unknown. To understand how an electroencephalograph is made, the brain may be pictured as a vast aggregation of electrical cells. When a million or so of these cells repeatedly discharge together the rhythm of their discharge becomes measurable in frequency (speed) and strength (amplitude or voltage). By the use of an amplifying apparatus these currents can be picked up, recorded graphically on paper, measured for pattern, and studied for their significance (see figure 11). Picking up these "waves" from the brain is a relatively simple procedure for the adult patient. Electrodes (small wires with a metal button or needle on the end) are pasted onto the scalp or inserted just under the skin through a part in the hair. The electrodes are moistened with a special salt jelly spread on the button to diminish the electrical resistance of the skin. The electrodes pick up the electrical currents generated by the brain; no current passes in the other direction from the machine to the patient. Each electrode is placed in a specific area of the scalp which in turn is recorded by a specific needle or pen on the moving graph (see figure 12). Generally, the number of electrodes used is eight.

The actual voltage or strength of these electrical currents is, of course, exceedingly small. The lighting system of a house is 110 volts. The electrical currents produced by the brain are measured in terms of microvolts. One microvolt is equivalent to one-millionth of a volt. Ten to 50 microvolts will usually cover

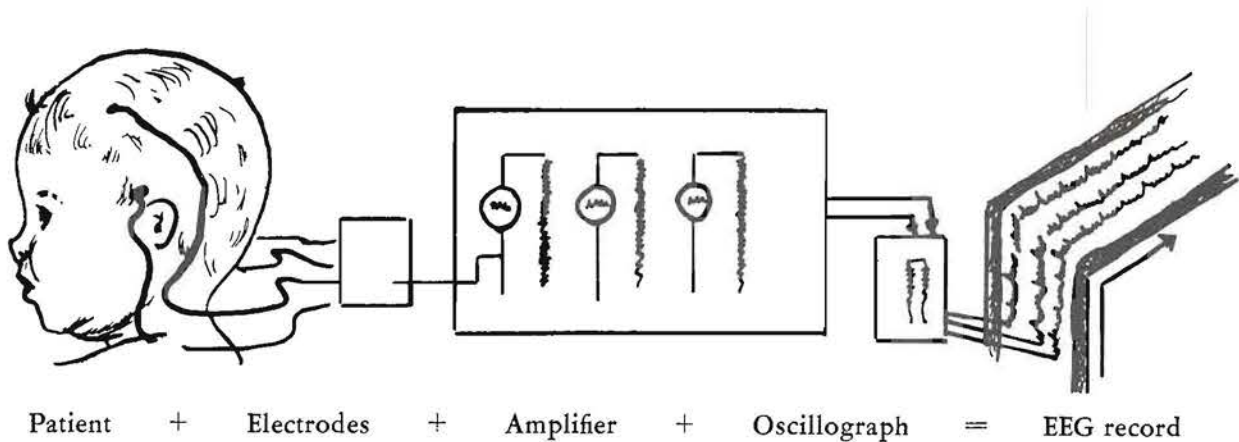


Figure 11.—COMPONENTS NEEDED FOR A RECORD OF CEREBRAL ELECTRICAL ACTIVITIES.

the range of the waves recorded by EEG machines. The greater the height—or amplitude of the wave, the greater the voltage (2).

If you move a pencil regularly up and down on a paper that is being drawn steadily from right to left, the result will be a regular series of curves. If at the same time the paper is moving up and down, another series of curves will be added to the line drawn. If the table is

shaking, the vibration will be added to the line as a ripple. There will then be three components integrated in the wavy line, which will begin to look something like an EEG record. The line gives a coded or conventional record of the various frequencies and amplitudes of various physical movements. An actual EEG record, however, contains as many as 20 to 30 significant components woven together from tens

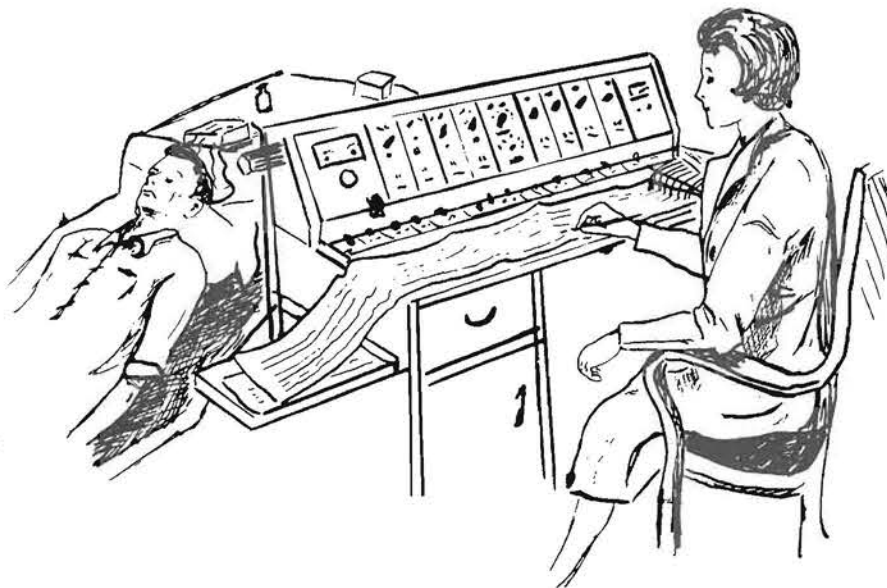


Figure 12.—TAKING AN ELECTROENCEPHALOGRAM—EEG.

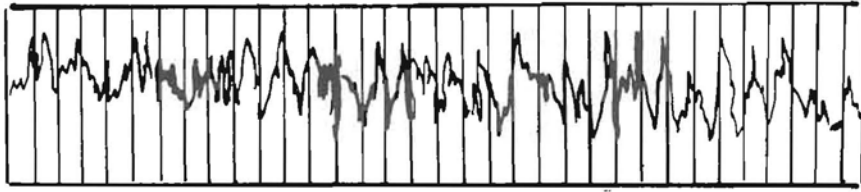


Figure 13.—EEG RECORD OF NORMAL ADULT ILLUSTRATING THE IRREGULAR DOMINANT RHYTHM OF 10 CYCLES PER SECOND.

Courtesy of Dr. R. Sedgwick, M.D., University of Southern California School of Medicine.

of thousands of impulses (1).

The record of a normal adult shows a fluctuating voltage recordable from the outer surface of the head as an irregular pulsation that usually has a dominant rhythm of 10 cycles per second (see figure 13). It would appear somewhat like the drawing below. Differences in patterns do occur which are more or less characteristic for each individual.

There are many variables in the use of the electroencephalogram. These include the sensitivity of the machine itself, how many channels are used to record (usually eight) and the time of the recording. Another major variable is the interpretation of the record. There are so many different ways to read these records that we can almost classify electroencephalographers into three classifications: (1) radicals, (2) moderates, and (3) conservatives. Some will

see an abnormality in almost every record they read; others seldom find any abnormality. The fact that no standards of normality and abnormality have been developed presents a real difficulty. There are some records of course in which there is 100 percent agreement; but too often when a definite opinion is needed, conflicting readings are offered.

The first source of variation is the patient himself. Unfortunately, this variability is most marked in children.

At birth, and for some months after, the main feature of the EEG is still irregular delta rhythms (see figure 14). The more passive and somnolent the infant, the more prominent are the delta rhythms. Even in the first few days of life there is a marked difference between the sleeping and waking patterns of the EEG. Changes of frequency are correlated best with

Brain wave patterns at various age levels

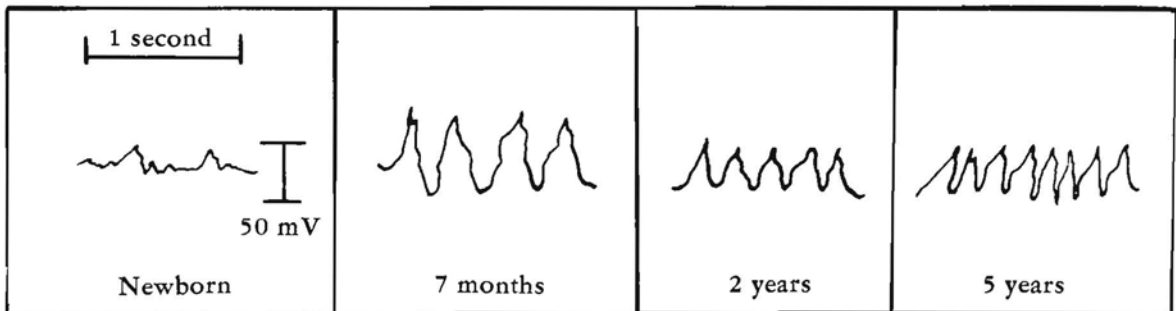


Figure 14.—ILLUSTRATING THE MATURING PATTERN OF BRAIN WAVES.

Courtesy of Dr. R. Sedgwick.



brain weight, changes of amplitude with the number of active neurons in the first few months and with skull thickness thereafter (1).

Figure 14 shows a flat, rather poorly organized record with just a few swiggles off the base line in the newborn. But, at 7 months of age the record is beginning to develop some recognizable slow waves. At 2 and 5 years of age, the graph shows an increase in frequency and variety. As the child matures you see less of this high voltage—slow wave abnormality. These changes in the electroencephalogram caused by a child's maturation make it difficult to interpret the record. One must take into consideration the maturation of the child in evaluating the EEG tracings. Somewhere between the ages of 9 and 16 years, a child's EEG reading begins to resemble an adult's.

The alpha rhythm (figure 15) is the most easily recognizable rhythm. It begins to become

apparent between 9 and 16 and is an 8- to 13-cycle-per-second rhythm, which is best defined or seen in the posterior parts of the brain. It is abolished when the eyes are opened, but is quite well defined when the eyes are closed. In other words, stimulation will cause blocking of the alpha rhythm. The alpha wave is the chief phenomenon of the EEG. The alpha rhythms will vary according to the state of awareness of the child—whether in a light or deep sleep. Many abnormalities are brought out in sleep.

The beta rhythm (figure 15) has an average frequency of about 25 waves per second; the delta waves have a duration of as long as one-sixth of a second (3).

In 1946, it was shown that the EEG pattern could be influenced by subjecting the brain to rhythmic stimulation. After World War II, an electronic stroboscope was used in this experiment as this instrument can be calibrated

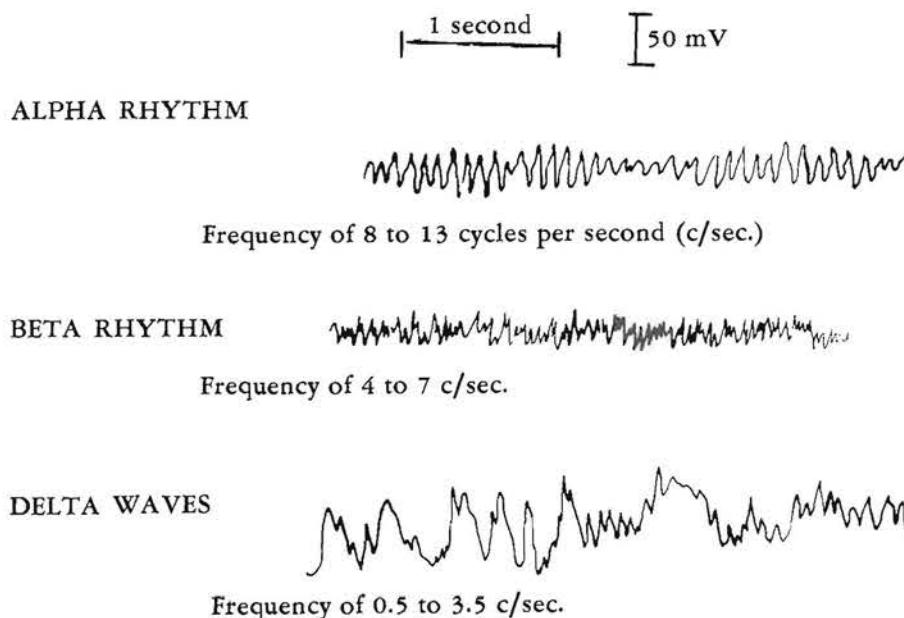


Figure 15.—CHIEF TYPES OF WAVES THAT MAY APPEAR IN THE HUMAN ELECTROENCEPHALOGRAM. THE FREQUENCY OF RHYTHM (THE NUMBER OF TIMES A CYCLE OF ITS PATTERN IS REPEATED IN A SECOND) IS MORE SIGNIFICANT THAN ITS AMPLITUDE OR VOLTAGE (mV).

From—"The EEG and Its Use In Psychology". Amer. Journal of Psychology, Vol. 51, No. 4, October 1938, pp. 737-750.

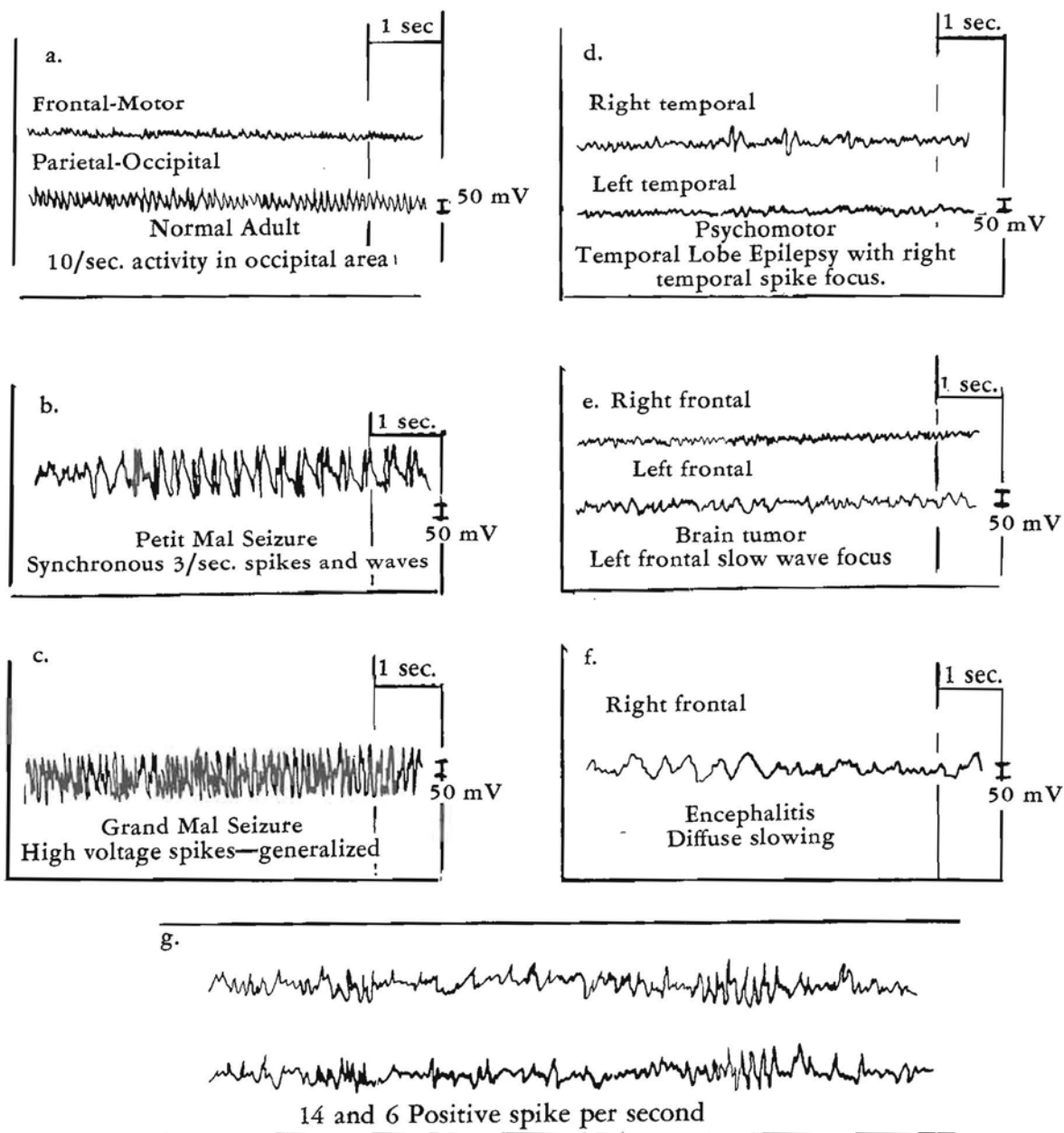


Figure 16.—NORMAL AND ABNORMAL TRACINGS.

R. Sedgwick, M.D., and Merck Manual of Diagnosis & Therapy, 10th edition. Merck, Sharpe & Dohme Research Laboratories.

in fractions of a cycle per second, using a very short brilliant flash, without varying the duration of frequency (*I*).

Should the child have a blood clot over the right side of the brain, the electrodes would

record suppressed impulses on that side while the record of the left-sided electrodes would show relatively well-developed rhythms. Should there be a clot on both sides, little activity would be recorded by the EEG.



A child suffering from encephalitis would show a diffuse slow record during the acute states. As he recovers from the illness, he has fewer slow waves, although he may not have normal rhythm.

Brain tumors, abscesses, clots, and focal injury may all give rise to focal EEG abnormality (see figure 16).

The technique for bringing out abnormalities through the EEG is termed an activating procedure. There are many activating procedures, but with the use of each there is a risk of introducing another variable in the record and thus making the correct interpretation even more difficult. In addition to sleep, hyperventilation can be called an "activating procedure." Whereas sleep brings psychomotor epilepsy to the fore, hyperventilation will almost routinely bring out the brain wave abnormality of petit mal epilepsy. Certain drugs, mainly Metrazol, are also considered activating agents.

Dr. Wilder Penfield of McGill University, Montreal, Canada, whose pioneering neurosurgery has relieved the suffering of hundreds of epileptics, used an electrode as an activating agent. During the 1920's and 1930's, Dr. Penfield devoted his life to epilepsy research which at that time was regarded by the medical profession as fruitless. He believed that epilepsy was not a disease, but a symptom of something awry in the brain. His persistent investigations showed that epilepsy was literally an electrical explosion, brought on by exceptionally heavy charges accumulating frequently in a damaged part of the brain. The damage in more than half of his patients was attributable to inadequate oxygenation or traumatic brain compression at birth. Dr. Penfield discovered that when certain parts of the brain were touched with the electrodes he could cause a leg to jump, or an eye to wink. He soon found that vivid recall of an incident that had happened to the patient years ago could be elicited by activating a particular portion of the brain (10).

The wizardry of medical electronics has made continued progress in recent years with the development of a miniature EEG unit by research workers at the Langley-Porter Neuropsychiatric Institute, University of California

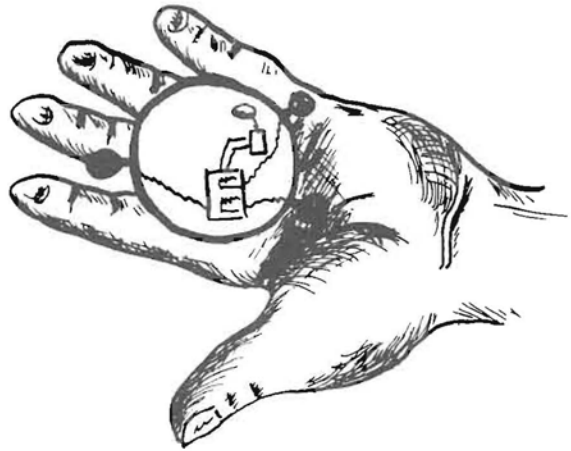


Figure 17.—WATCH SIZE EEG PERFECTED AT LANGLEY-PORTER, SAN FRANCISCO, CALIFORNIA—1962.

School of Medicine. This tiny EEG unit (see figure 17) has been used to record brain waves of disturbed children. This unit is approximately the size of a watch and weighs 10 grams. The three round electrodes are attached to the scalp with tape. The child is completely free from wires or heavy equipment and thus is able to participate in normal activities. The compact device contains batteries good for 5 hours continuous recording. The batteries power a tiny, self-contained radio transmitter that amplifies the electrical impulses produced by the brain and records them on tape via an inkwriter.

### *What is an epileptic seizure?*

Epilepsy is not a disease but a symptom of an abnormal electrochemical occurrence in the brain. Seizures may occur in any individual and they may have their onset at any age. Different individuals have different levels of sensitivity toward having a convulsion or seizure. Some of the factors responsible: (1) abnormalities in development of the brain (or destruction of its structure) and coverings; (2) abnormalities in the functioning of the cells, and consequent increase in their convulsive responsiveness, and (3) abnormalities in the body other than the brain.

Little is known about the mechanism of an

✓  
epileptic seizure and its many physiologic symptoms, but we do know that an important function of the brain is to change energy derived from sugar and oxygen circulating in the blood into nervous energy. A seizure results from the failure of the brain to limit and control the time and place of its release of this nervous energy. The usual lethargy shown by most children after an attack indicates that all this store of excess nervous energy has been exhausted. Other known causes are the lack of, or decrease of, oxygen or blood to the brain; ingestion or inhaling poisonous substances; electrolyte imbalance; hypoglycemia (low blood sugar); various types of central nervous system infections; or brain tumor.

Children whose difficulties are due to maldevelopment or to destruction of the brain, and those children who suffer seizures for unknown reasons have in common the fact that their seizures are due to a sudden disorderly discharge of the brain cells. This sudden spread of an explosive discharge of electrical energy from a focal point affects the functions of otherwise normally discharging neurons. The type of seizure differs widely depending upon the focal point of discharge and on the speed with which it travels to distant brain areas.

Rarely, when taking an EEG, is a child experiencing a clinical seizure. In other words, the machine is recording interseizure voltages from the brain, and on occasion the abnormality may not be picked up. Sometimes the paroxysmal mass discharge of neurons may be picked up on the electroencephalogram and yet, the patient may have no clinical symptomatology. The question arises then, does he or does he not have epilepsy? Conversely, if the child has a seizure, and yet he consistently has a normal electroencephalogram, can he be diagnosed as having epilepsy? Some doctors feel that the diagnosis should be based on whether or not the child has an active clinical seizure. Seizures can be roughly classified into major groups. The most familiar is the grand mal seizure.

### *Grand mal*

The grand mal convulsion (figure 16) results from the spread of electrical activity through-

out the entire brain. It is a generalized seizure, the essential features of which are loss of consciousness and tonic (stiffening), then clonic (jerking) movements of the whole body. The attack usually begins with an aura or warning characterized by some unusual sensations—unpleasant smell, flashing colored lights, abnormal hearing, tingling of some part of the body, loss of speech or some other complaint. This aura is then followed by muscular contractions. The head is pulled to the side and held stiff, the features are distorted, the pupils dilated, and the rigidity of the muscles prevents respiration. This is followed by jerking movements of the entire body. There is also a chewing motion and a blue discoloration of the face due to decreased respiration. The patient froths at the mouth, and sometimes bites his tongue quite severely. Occasionally, loss of bladder control accompanies these attacks. This stage lasts from 30 seconds to 5 minutes or longer. When the patient regains consciousness, he may be drowsy, exhausted, and depressed, have a headache, or be unable to function clearly for a variable period of time, but he has no recollection of the attack. In rare cases the patient recovers feeling unusually fit and fresh.

The EEG record may show hypersynchrony throughout the entire cortex if the patient is having a grand mal seizure during the examination, or even a subclinical interseizure discharge. This usually has the same connotation as the petit mal three-cycle-per-second; that is, it is not ordinarily a concomitant of disease or injury to the brain.

### *Petit mal*

This type of seizure (figure 16) is characterized by a brief interruption of consciousness. It may pass almost unnoticed with the patient staring or having a lapse of full awareness for a few moments (5 to 30 seconds is usual). The patient rarely falls, although the head may drop forward or turn to one side and the eyes are veiled and expressionless for a few seconds. A patient may stop in the middle of a sentence, stare vacantly, blink several times, and then continue speaking. At times his train of

thought might be interrupted and, when he resumes speaking, his completion of the sentence will have no relation to his original idea. A little rhythmic twitching of eyelids or eyebrows may occur. Rarely does the twitching involve the arms and shoulders. At times the patient may drop what he has in his hands or fall to the floor and get right up without realizing he has just had a seizure.

The EEG records show the classical three-cycle-per-second Dart and Dome wave, which is called the petit mal record. However, finding this type of record in a patient does not mean that he has petit mal; he may have grand mal only.

This type of record also points to the so-called idiopathic seizure; that is, the seizure not due to disease and possibly having familial determinants rather than the seizure due to disease or brain injury.

### *Psychomotor*

This is the most complex type of seizure. The abnormally discharging cells act upon the mental process as well as upon muscles. During these seizures the patient goes through motions which appear to be purposeful but which are not relevant to the situation. These may be chewing motions, smacking of the lips, or buttoning or unbuttoning clothes. The patient gives the impression of being intoxicated by wandering around. On rare occasions, these spells take the form of rage states. There is no memory of what took place when the seizure is over.

The EEG record may show a spike focus primarily located in the temporal lobe (see figure 16).

### *Focal*

Focal seizures are attacks which start in one part of the body and may remain entirely limited to that part. Often the attacks occur without loss of consciousness. However, there may be a spread of the orderly discharge. The seizure may start in the toes of one foot or the fingers of one hand, or in one corner of the mouth. Suddenly the affected part trembles

violently, or simply feels numb. As more and more neurons become affected, the seizure spreads. It may stop at any time or, in a few seconds or minutes, cross to the other side of the body. This terminates then in unconsciousness and a grand mal attack.

### *The 14 and 6 positive spike record*

Children with this type of EEG abnormality (figure 16) are said to have increased incidence of recurrent headache, abdominal pain, and behavior disorders.

### *Myoclonic seizures*

These are brief, nonpatterned motor seizures often symptomatic of serious diffuse brain disease. A common EEG finding in this particular problem is called "hypsarrhythmia." It has only recently been recognized and shows a wild slow wave and spike wave discharge of high voltage. Treatment of this group has been more difficult than other types.

### *Summary*

The electroencephalograph is an instrument which records the electrical discharges of the brain cells. It can record millions of cells discharging in synchrony, amplify the voltages some 1 to 2 million times and record on a graph by use of moving pens. There are many variables in the use of this machine. Therefore, the electroencephalogram of a patient can vary from hour to hour, day to day, depending upon his diet, the amount of rest, whether he has been ill recently, or what drug or drugs he may be taking. Successive records of this patient may show marked variability. The doctors must interpret with caution any certain statement of diagnosis or prognosis on the basis of one record. The EEG is simply an ancillary tool in neurologic diagnosis. It does not make a definite record of whether or not the child is retarded. The EEG neither rules in brain damage or rules it out. It is, however, a valuable additional diagnostic procedure, but should never be used as a single diagnostic tool.

Each of the seizure problems has its own specific therapy. These seizures form an important factor in mental retardation by reason of their direct effect on mental performance and possibly through the actual production of brain damage by the seizure. Knowledge of the nature and spread of the epileptic discharge is being advanced through electrophysiological and electrochemical studies of neuronal activity in the brain (9).

It is stated by Yannet that approximately 15 percent of institutionalized retardates may have convulsions (11). In Tarjan's studies, epileptic seizures were present in 536 of 2,000 retarded patients (9).

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# OPHTHALMOLOGIC PROBLEMS

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A GREAT NUMBER of mentally retarded persons have visual problems. In fact, 30 per cent of the children seen at the Child Development Clinic at Childrens Hospital of Los Angeles have ophthalmologic difficulties. Some years ago, Dr. Margaret Jones, formerly of the cerebral palsy clinic at the same hospital, made this statement, "If you can take care of the other handicaps, then we can concentrate on the main problem." The usual defect is strabismus, or cross-eye. This is a twofold condition. When it is corrected, the general appearance of the child is improved. But even more important, the eyes will be physiologically more efficient. There are many misconceptions about strabismus; generically, this term covers cross-eye, wall eye, and vertical eye. Most people assume that one eye crosses—either the left crosses to the right or vice versa. But this condition is not a muscular one; it is not due to weak eye muscles or strong eye muscles. Strabismus is due to the fact that one eye or both eyes do not focus properly.

## *Vision*

Vision is dependent upon several factors; the bulb of the eye or the eyeball, the optic nerve, and the visual center in the brain. In addition to these essential organs, there are accessory organs necessary for the protection and functioning of the eyeball. These are the eyebrow, eyelid, eyelashes, conjunctiva, lacrimal apparatus, muscles of the eyeball, and the fascia bulbi (see figure 18).

**Eyebrows**—Interlaced beneath these two arches of thickened skin are the muscles of expression.

**Eyelids**—The eyelids are two fibromuscular curtains that help protect the eyeball from

bright light and foreign objects, cover the eye during sleep, and spread lubricating secretions of the eye over the surface of the eyeball.

**Eyelashes**—The eyelashes project from the margin of each eyelid. The follicles of the lashes receive a lubricating fluid from the sebaceous glands which open along the margin of the eyelids.

**Conjunctiva**—The conjunctiva is the mucous membrane which lines the eyelids and is reflected over the forepart of the eyeball.

**Lacrimal apparatus**—The lacrimal apparatus consists of: (1) the lacrimal gland, (2) the lacrimal ducts, (3) the lacrimal sac, and (4) the nasolacrimal duct. The lacrimal gland secretes tears to keep the eyeball moist and to wash away any foreign objects on the eyeball. Every time you blink, the eye is washed. Ordinarily this secretion is evaporated or carried away by the nasolacrimal duct as fast as formed; but under certain conditions or circumstances, the secretion of the lacrimal gland exceeds the drainage power of the nasolacrimal duct, and the fluid accumulates between the lids and overflows down the cheeks as tears. The lacrimal sac is formed by the union of the duct from each eyelid and drains into the nasolacrimal duct.

**Eye muscles**—The six muscles of the eye control the sideward motion of each eye. In addition there is a muscle in the upper eyelid to control the eye's up-and-down motion. This muscle is the Levator palpebrae. All of the eye muscles originate at the back of the orbit and come forward to different areas of attachment at the anterior part of the eyeball. Through centers in



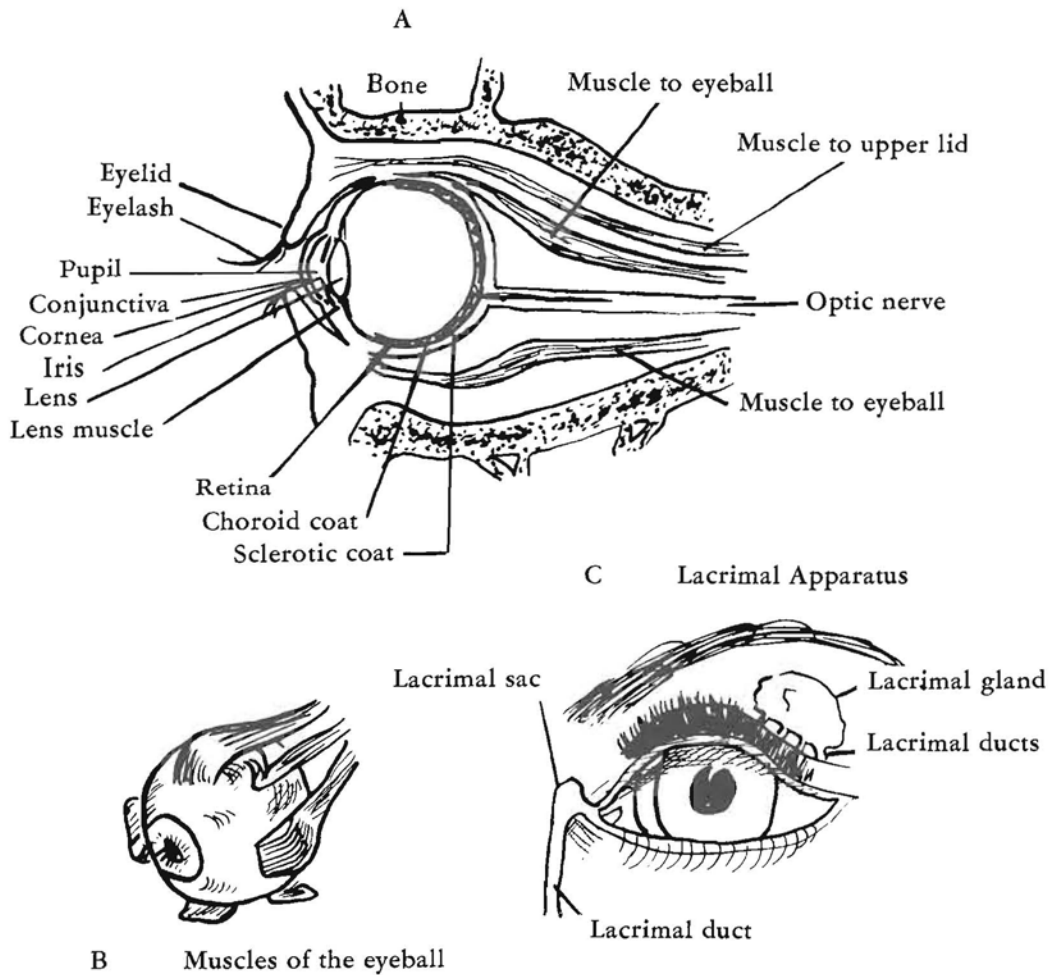


Figure 18.—A ILLUSTRATES THE SAGITTAL SECTION OF THE ORBIT AND EYELIDS, B—MUSCLES OF THE EYEBALL, AND C—THE LACRIMAL APPARATUS.

Cates' Primary Anatomy by J. V. Basmajian, M.D.—pp. 333.

the brain, these 14 muscles work together to guide the two eyes to focus as a unit on various objects near and far.

**Fascia bulbi**—This is the connective tissue surrounding the eyeball.

**Eyeball**—The eyeball is a hollow sphere measuring about an inch in diameter. It is covered by a tough lining, the *sclera*, which is white in color. In front, this lining becomes more delicate and completely

transparent and is known as the *cornea*. Just behind the cornea lies a cavity filled with a fluid called *aqueous humor*. Behind the aqueous humor and in the center of the pupil is the *crystalline lens* of the eye. The lens is partly covered by a circular membrane known as the *iris*. It is this membrane which gives the individual coloring of the eyes. The dark opening in the center of the iris is the *pupil*. This is the point through which light rays are transmitted



to the lens. The lens is made up of a substance which can be changed in shape by the action of *ciliary muscles* which surround its outer edge. Behind the lens is a large chamber filled with a gelatinous material called *vitreous humor*. The light that enters the eye through the lens is focused on the rear wall of the eyeball. This wall is covered by the *retina*, which records the

image of the object seen. The normal eye can focus an image properly on the retina no matter whether the object is near or far. This ability is termed *accommodation*. The retina contains many millions of tiny organs which are sensitive to the effect of light rays. The *rods* enable us to see in dim light, but they do not form a very clear picture of the outside world. This is the

### Three dimensional stereoscopic sight

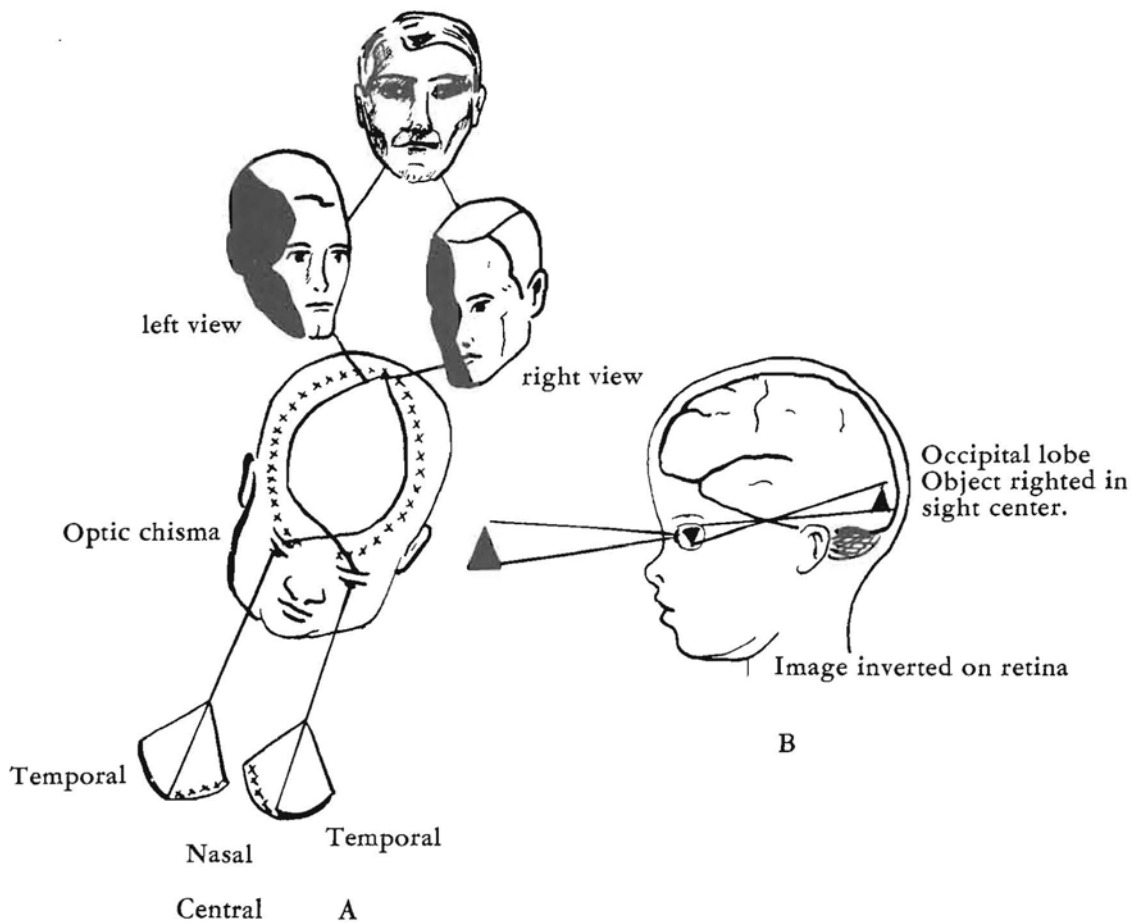


Figure 19.—ILLUSTRATING VISUAL FIELDS AND INTERPRETATION OF WHAT THE BRAIN RECEIVES IN A. IN B THE INVERTED IMAGE IS PRODUCED ON THE RETINA OF THE EYE AND A NORMALLY FUNCTIONING BRAIN RIGHTS THE IMAGE OF US IN THE SIGHT CENTER.

responsibility of the *cones*. They allow us to see color and fine details. The rods contain a substance called *visual purple*. When an abnormality occurs here, the individual cannot see well in dim light and has "night blindness."

**Optic nerves and visual centers of the brain—**The eyes can be likened to a camera—each eye takes a picture, then the brain puts the two pictures together. The left half of our brain receives the nerve fibers from the left half of both eyes. The right half of the brain receives the fibers from the right half of both eyes. The brain then fuses these two pictures, which are different. By fusing the two pictures the brain conveys depth perception, which enables us to judge the distance of various objects (figure 19).

Under normal conditions, it takes us several hundredths of a second to see an object. If the object is difficult to see because of lack of light and because it is not contrasted with its background, it may take longer. When the retina receives an image from the reflected light of an object, the image remains in the eye for approximately a 10th of a second after the light is removed. Our ability to see depends upon both the amount of light present and the contrast of the object with its surroundings.

In every 1,000 babies born each year, 15 will have the problem of having a third dimension somewhat like a stereo camera (see figure 19). This condition is mistakenly called a muscular defect of the eyes. It does not necessarily occur at birth; in fact only a small percentage have it at birth. But it does appear in the first few years of life, usually between 2 and 4 years of age. Whether the child is normal or mentally retarded, the problem still exists. The brain, for reasons that are at present unknown, will pick up the vision on one side, but it seems to drop a curtain over the other side. This causes the eye to drift inward, *esotropia*, or may cause it to drift outward, *exotropia*. In less than two percent, one eye drifts up and the other down.

For example: (1) if the child fixes with the left eye, the right one tends to drift in or cross; (2) if the child fixes his point of vision to the

left, the right eye may go in and slightly up; (3) the same child may fix with his right eye and cause the left to drift in. The particular eye involvement depends on where the attention point may be. On occasion you will have a condition called wall eye or *divergent strabismus*. Here, the eyes turn out toward the temples. When the child fixes with one eye, the other drifts outwards. The eyes actually move in opposition to each other.

The treatment for this condition is a long range project. First, special medicines are dropped into the eye to dilate the pupils for examination. This is necessary to be sure that the seeing structure, or the back of the eye, is normal. In the actual process of seeing a beam of light passes through the lens and is turned upside down as well as being reversed from right to left. After passing through the lens, light traverses a large spherical cavity that makes up the bulk of the eye. This cavity is filled with a clear liquid through which light passes easily. The image is focused on the retina. From the retina, the image is carried to the brain for interpretation (figure 19).

### *Refractive errors*

Assuming that this seeing structure is normal, the child is checked to see if glasses will help his focusing ability. Twenty-five percent of cross-eyed children are very farsighted. When they receive glasses, their focusing ability relaxes and the eyes will be straight while wearing the glasses (see figure 20). Farsightedness, or *hypermetropia*, is a condition in which rays of light from near objects do not converge soon enough and are brought to a focus behind the retina. The eye must accommodate slightly for distant objects and overaccommodate for near objects. Hypermetropia is usually caused by a flattened condition of the lens or cornea, or an eyeball that is too shallow. The condition requires a convex lens to concentrate and focus the rays more quickly.

In figure 20 the parallel lines indicate light rays entering the eye. X is the point of convergence or focus.

In (A), the rays are brought to a focus directly on the retina in normal sight.

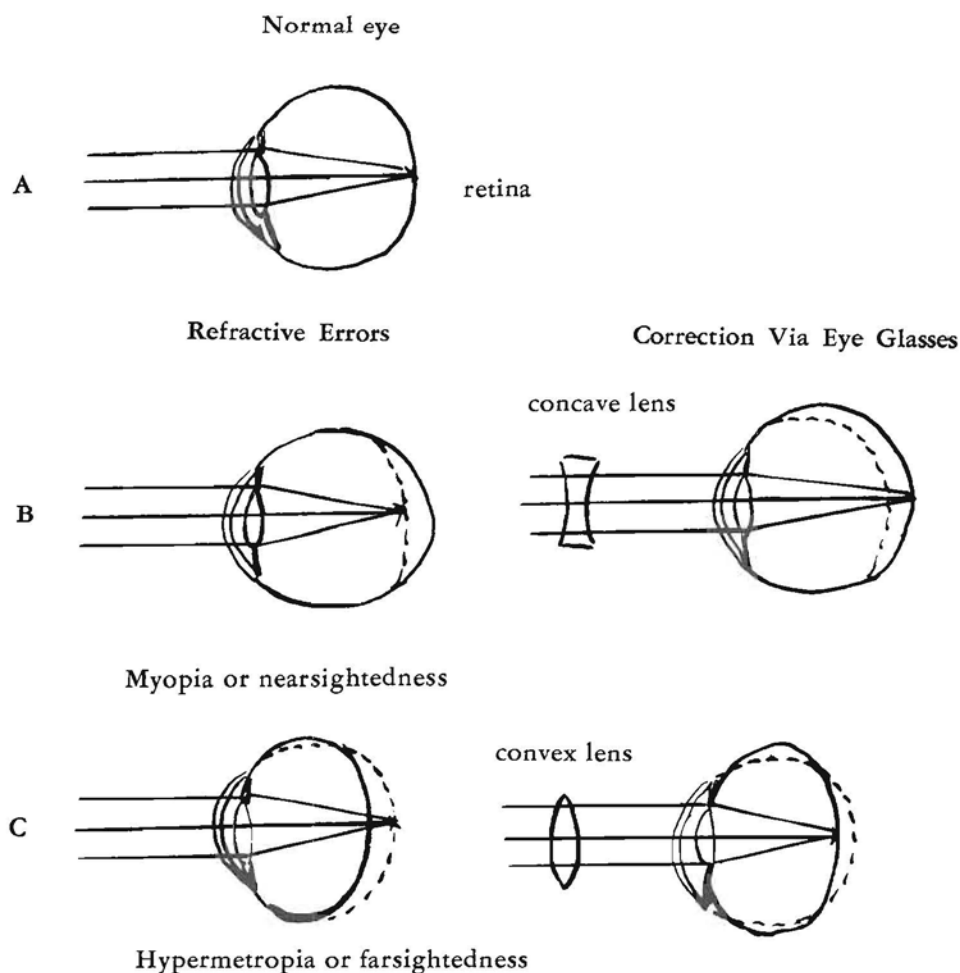


Figure 20.—SCHEME TO DEMONSTRATE THE FOCUSING OF PARALLEL RAYS OF LIGHT (DISTANT VISION) AND DIVERGING RAYS (NEAR VISION) BY THE CORNEA AND LENS IN NORMAL AND ABNORMAL SIGHT.

In (B), the nearsighted eye, they come to a focus in front of the retina. The condition is termed *myopia*. A concave lens causes the parallel rays of light to diverge before they converge and focus on the retina. This condition results from an eyeball that has too much depth or from a cornea or lens that is too convex.

In (C), the opposite occurs. The eyeball is too shallow causing the rays to focus behind the retina. This is farsightedness or *hypermetropia*. A convex lens is used for correction of this condition.

Less than 2 percent of the human race have perfectly normal eyes. We are born with a short eyeball from front to back, meaning we are farsighted. As the child grows, the eye grows in length and size. An infant's eye is approximately two-thirds the size of an adult eye. In adolescence, around the age of 14 years, the eye is approximately an inch long. Since our eyes do not come off an assembly line, there will be a marked difference in size. Some are a little shorter, requiring the need for glasses to assist them in reading and close work, while others are a little longer, requiring

glasses for distant objects.

Twenty percent of the population are or become nearsighted. The eyeball grows too long. The cause is unknown. The condition can exist at birth, but is rare. *Congenital myopia* is found in some handicapped children, but the majority of children having this condition acquire it in preadolescence and adolescence. This coincides with an increase in hormonal function seen in the sudden spurt in height. Vision is clear close up, but blurred at a distance. Glasses aid in improving the vision but do not cure the handicap. In school the child is unable to see the blackboard without the aid of glasses and may be considered retarded due to lack of attention or continued mistakes in copying a lesson. Again, whether the child is retarded or normal, nearsightedness has nothing to do with the use of the eyes. In the Orient where 99 percent of the population is illiterate, 30 percent of the people are myopic in contrast to 20 percent in Western Europe and America.

The other visual problems seen in children are surgical in the sense that the eyes can be realigned; we cannot, however, correct the way rays are deposited onto the retina. In retarded children there are several problems that occur when vision is being strengthened in one eye. Where the child is using one eye and suppressing the vision in the other, the good eye must be covered in order to make the other eye focus. This is a hardship on normal children and the problem is intensified when the child is retarded. This procedure should be started before the child reaches school age. Oddly enough, one of the greatest helps in strengthening lazy eyes is television.

Usually, vision can be tested accurately at the age of 4½ years. Before that age, the doctor can only surmise. A combination of glasses and surgery is used to help many cases. And a child is followed for many years after surgery and/or glasses are fitted. Together with this combination exercises are used, but one cannot exercise eyes as they do biceps in the gym. The eye is controlled by the brain and one cannot exercise the brain. Instead, a technique called *orthoptics* is used. An orthoptic technician trains the child to look into an instrument much like grandmother's old-fashioned stereopticon that one could look in and see three dimensional

scenes. This machine helps the child learn to use his eyes together. He is asked to look with first one eye, then the other, then both until he is able to see the objects pictured within at the proper perspective and third dimension vision is attained. On one side a bird may be pictured; on the other a cage. When he looks with both eyes, the child should be able to see the bird in the cage. In dealing with retarded children, this procedure is extremely complex, and orthoptics is not always possible. Refraction is attempted and glasses fitted when indicated.

Another problem seen in retarded children is *cataracts*. There are many types of cataracts caused by a variety of conditions; some are congenital. If an expectant mother has rubella (German measles) during her first trimester, there is a good chance that after the child is born, he will exhibit the rubella syndrome which consists of deafness, retardation, a cardiac condition, and congenital cataracts. Some statisticians say that approximately 12 percent of expectant mothers who have rubella in the first trimester will have a child with such defects. Other authorities say that the percentage is even higher.

### *What is a cataract?*

A cataract is not a film, a growth, or a tumor. It is a clouding of the crystalline lens of the eye. With a cataract, there may be blood-vessel abnormality as well. Thus the affected child can distinguish light and dark only. In a few instances the child can see around the clouded part of the eye.

Also associated with cataracts are other diseases or congenital defects whose causes are largely unknown. In Marfan's syndrome, the child will be extremely tall and thin with long fingers and toes (also known as spider hands and feet) and have cataracts. The lenses are not completely cloudy, but dislocated. There is an area in which the child has vision but only with very thick glasses to assist him. He cannot see through the lens. There are case histories of this condition being reproduced from generation to generation.

Congenital cataracts are different from

adult cataracts or senile cataracts. There is a strong possibility that everyone reaching the age of 60 years will have some clouding of the lens of the eye. The condition may start at 60 and progress through the years until vision is clouded and requires surgery. In children the cataracts are usually relatively dense and, unless surgery is performed early, the child, in trying to adjust his vision, will develop nystagmus. This condition is characterized by a jerky movement of the eyes. Once nystagmus develops, it is certain that the child will never have better than 20-200 vision after cataract surgery. This amount of vision allows the child to get around and take care of himself, but he is severely handicapped. If surgery is performed before nystagmus develops, it is likely that better than 20-200 vision can be obtained.

One of the diseases causing cataracts is *galactosemia*. This type of cataract is not very dense. Ordinarily a cataract involves the entire lens. A surgical technique used in some cases is an iridectomy. This is a partial removal of the upper part of the iris. The child sees through the area above the cataract. Another surgical procedure was to cut away a portion of the lens in successive operations. The procedure now is to remove the lens completely. When the lens is removed the patient is no longer able to focus. Cataract glasses (bifocals) which are extremely thick and magnify are prescribed.

In the *Sturge-Weber syndrome* (cerebral angiomas) the cataract is due to enlarged blood vessels and is called telangiectasia. Mental retardation may be present in this condition which can be identified by a red birthmark on one side of the face. Unfortunately the vessels in the iris may block the drainage angle of the eye resulting in glaucoma which is a condition characterized by an increase in the pressure within the eye.

*Toxoplasmosis* is an infection by lower organisms in which there is an inflammation in the retina (this is the film of the camera—the seeing part of the eye) which may result in very poor vision. The same process may occur in the child's brain, causing retardation. The vector spreading toxoplasmosis is unknown but

a contributing factor may be ingestion of raw meat such as mutton, hamburger, or bacon. It is a type of infection that may be passed from mother to child during the early part of pregnancy, similar to rubella.

*Francischetti syndrome* is characterized by a tumor composed of blood vessels (hemangioma). It is similar in appearance to the Sturge-Weber syndrome but does not have a true port wine stain with glaucoma on the affected side. This syndrome was identified and described by Dr. Francischetti, a professor of ophthalmology, at the University of Geneva. The affected patient has an unusually wide mouth (macrostomia) in addition to minute tumors growing on the eye and a notchlike defect in the eyelid. There are also extra appendages on one or both ears.

*Lowe's syndrome* is a relatively rare condition involving the eyes, kidneys, and brain from birth. The lenses are cloudy from cataracts and glaucoma is present. These children, unfortunately, do not develop well and their life span is short.

A cause of visual impairment that went unnoticed for many years was the use of too much oxygen in the care of premature babies. The condition that resulted was called *retrolental fibroplasia*. Fortunately for some children, the duration for their need of oxygen was short and their sight only partially impaired. Others were not so fortunate and became blind.

A problem seen in some retarded children is detachment of the retina. These children appear to see well in the morning but by evening they begin to fall over objects. During bedrest the retina flattens back onto the choroid by gravity. During the activities of the day the retina becomes partially or completely detached from the choroid. Diagnosis is made with the ophthalmoscope, which may reveal irregularities in the level of the retina, fold, tears, and black blood vessels. The prognosis in the average case is good if appropriate surgical treatment is given early. The patient is immobilized in bed; drops instilled; and the eyes bandaged pending complete evaluation (4).

*Ptoxis* (drooping of the eyelids) causes vis-



ual problems because the child cannot lift the eyelids. In order to see, it is necessary to lean the head back resulting, over a period of time, in lordosis. This then becomes an orthopedic problem as well; therefore, early treatment is a *must*. This condition may be familial in nature.

### *Summary*

Visual handicaps in a child should be corrected no matter how severe the mental handicap may seem. An increase of even 1 percent in vision may make a tremendous difference in a child's progress since children will make the most use of whatever vision they have. An adult who loses part of his vision may spend the rest of his life complaining about it, but a child just accepts it and uses what he can. In addition to giving the retarded child as much sight as possible, it is of cosmetic value to correct

obvious defects so that the child's appearance will be as normal as possible.

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# NEW DEVELOPMENTS IN GALACTOSEMIA

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THE STUDY AND TREATMENT of genetic diseases, especially those which are associated with mental retardation, are becoming increasingly important in pediatrics.

Genetics is the study of heredity—it is sometimes defined as the science that seeks to explain the similarities and differences that exist between organisms related by descent (1). The science of genetics is a branch of biology. In order to explore all of the complex reactions which are involved in the transmission of inherited characteristics through several generations, scientists must utilize the skills developed by such related sciences as chemistry, mathematics, and physics.

The word gene is used to designate each biochemical unit of heredity within a cell. From the brilliant intuitive concepts expressed by Sir Archibald Garrod in the early 1920's and by Professor Linus Pauling and his associates at the California Institute of Technology, improved methods of genetic and biochemical study are becoming available. Based upon such studies, the physician can develop logical procedures for patient management and useful sociological approaches to counseling.

From the scientist's point of view, the continuing discovery of "experiments of nature" provides models for expanding the current concepts of genetics. Among the genetic diseases now known, galactosemia serves as an excellent example of such an "experiment of nature" which has resulted in the development of new knowledge concerning galactose metabolism.

The disease has been known since 1908.

Individuals who have it cannot metabolize galactose. It was not, however, until 1940 that the disease was found to be due to an enzyme defect—an inborn error of metabolism. A group of biochemists in South America and several scientists in other countries demonstrated the normal pathways of galactose metabolism in bacteria, yeast, and certain animals in 1950. In the human, these pathways were clearly delineated a few years ago when Dr. Herman Kalckar and his colleagues at the National Institutes of Health discovered that the defective enzyme was galactose-uridyl transferase. In the process, methods of measurements were devised which are now applied as diagnostic tools. These tools have been utilized to establish the mode of transmission of the disease. Further studies of galactosemia undoubtedly will result in additional information concerning other knowledge of galactose metabolism.

## *Genetic aspects*

Research has shown that there are several different kinds of chromosomal abnormalities found in man. These include the presence of a single additional chromosome making a group of three instead of the normal pair. This grouping of three is called a *trisome*. The *monosome* is characterized by the absence of one chromosome of the pair that should be there. The absence of a part of a chromosome is called *deletion*. *Translocation* is the name for the ab-

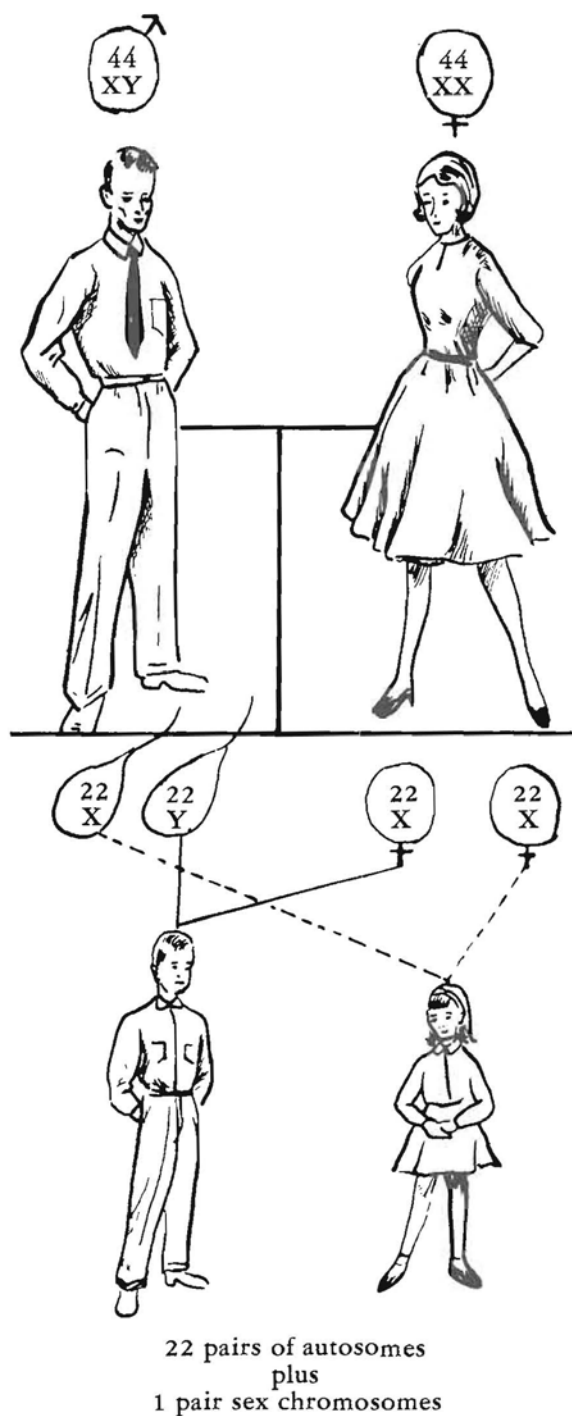


Figure 21.—THE XY METHOD OF SEX DETERMINATION BY CHROMOSOMES.

normality caused by a portion of one chromosome moving from where it should be to another chromosome where it should not be. The final abnormality, *triploidy*, occurs when an individual has half again as many chromosomes as he should have.

In man there are normally 22 pairs of chromosomes which are called autosomes. In addition there are the sex-determinants labeled X in the female and Y in the male for a total of 23 pairs or 46 chromosomes (see figure 21).

For centuries people were puzzled by the fact that certain hereditary characteristics appeared more frequently in males than in females; and yet, the males in each generation seemed to inherit the particular characteristic through their mothers and not through their fathers. The discovery of the chromosome mechanism of sex determination led to the clearing up of this mystery (2). There are 22 pairs of autosomes plus one pair of X sex chromosomes in the female and 22 autosomes plus one X and Y pair of sex chromosomes in the male. The total number of chromosomes is 46. When the female ovum develops (oogenesis) the chromosomes pair and the eggs and polar bodies all receive the same kind of chromosomes and one X chromosome. This is not the case in spermatogenesis (production of the male sperm) however, for one pair of chromosomes is unlike—the sex chromosomes XY. Half the sperm receive the 22 autosomes plus one Y chromosome while the other half receive 22 autosomes plus the X chromosome. Sex determination therefore, depends upon which fertilizes the egg. The sperm carrying the X chromosome is termed the female determinant while those carrying the Y chromosome are male-determining. Thus, when an egg unites with a sperm containing the X chromosome, an XX or female child is produced. When an egg unites with a Y chromosome sperm, an XY or male child is produced. Sex cannot be determined in advance due to the fact that only one sperm of the large quantity which is produced actually succeeds in uniting with the female ovum. By the same token, not every ovum that is produced becomes fertilized by a sperm cell.

There is no available explanation for most of the many abnormalities that occur in the

human race. In certain well-established hereditary diseases however, the cause is related to the presence of recessive or dominant genes located in the various chromosomes.

*Galactosemia* is such a condition and is caused by a recessive gene present in both parents. Both parents are clinically normal but carry the trait indicated by (g) in figure 22. The condition is not evident in the parents be-

cause the normal counterpart of the gene (G) is dominant. When both parents have a recessive galactosemic gene (g) the disease is produced in one out of four offspring. A person carrying a recessive gene and a dominant gene is identified as being a *heterozygote*. A *homozygote* carries a pair of like genes and either manifests the characteristics of disease, or is a normal noncarrier.

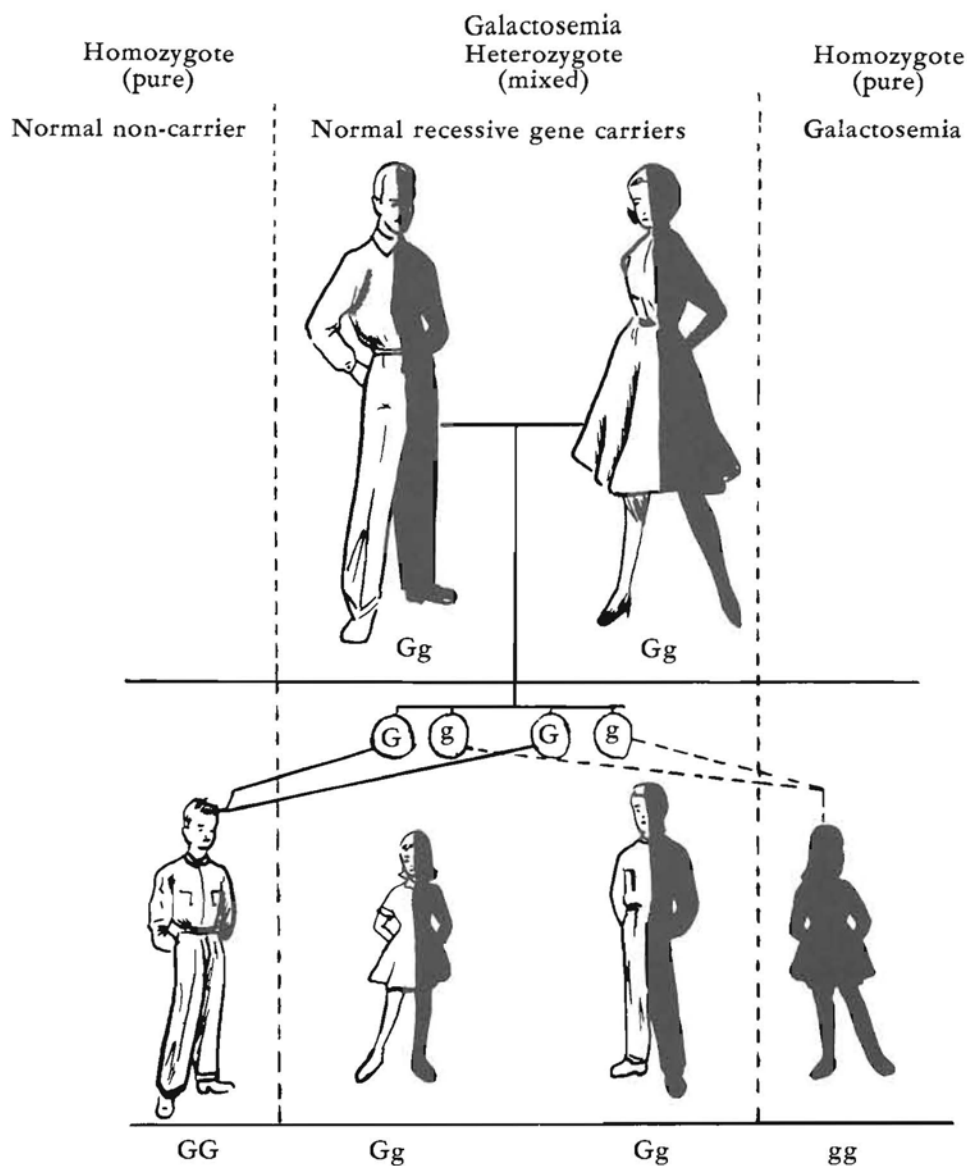


Figure 22.—GENETIC TRANSMISSION OF GALACTOSEMIA.

Clinically it was observed that affected children were born of parents who themselves did not demonstrate any abnormality. Therefore, it was suspected that the transmission of the defect was recessive. A dominant character would be seen vertically in a pedigree from one generation to the other, while recessives are limited to the immediate generation. The recessive theory was substantiated further by the appearance of the disease in siblings, but not in cousins. It was also noted that both boys and girls seemed to be equally affected, indicating the involvement of an autosome chromosomal defect rather than a sex chromosome abnormality.

Because of the rare occurrence of the disease, it was impossible to establish either of these points on a sound statistical basis. Establishment of the mode of transmission had to await the further evaluation of the fundamental defect.

In the past the *galactose tolerance test* was used in diagnosis. This direct approach is a laboratory procedure based on the concept that the essential feature of the disease is an inability to metabolize galactose in the normal manner. In the test, the individual is given an oral dose of galactose which is sufficient to put the suspected enzyme system under stress. In the case of galactosemia this is the transferase system. Judgement of the physiological response is made on the basis of the speed with which galactose disappears from the blood stream. In the normal individual, the percentage of sugar in the blood goes up approximately a third of the way seen in a galactosemic, and by 3 hours drops to normal. In studying family members of a galactosemic child, it was found that many have responses which are intermediate between the galactosemic and the normal. It was therefore impossible to make a diagnosis on the basis of the galactose tolerance test alone.

## *Metabolism*

It is of academic interest to learn how galactose is metabolized by a normal individual and it is of extreme medical importance to understand how a galactosemic can omit this sugar from his diet.

As an infant, the individual normally consumes galactose (a milk sugar) in his first feeding. Throughout life, galactose continues to be present in his food intake—in the lactose of all milk products.

In arriving at a scheme for the metabolism of galactose, two major points must be resolved. First, how is dietary galactose used to provide energy; second, since this sugar is an essential part of the structure of a variety of important compounds in the functioning of the body, can it be supplied and synthesized within the body itself once the sugar is no longer available from outside dietary sources.

The weight of evidence at the present time is that dietary galactose can provide energy only by conversion to glucose 1-phosphate, which is an intermediate in the pathway of glucose metabolism. When comparing the sugar molecules of glucose (normal sugar) and galactose, the only difference noted is in the position of the hydroxyl group on Carbon No. 4. Otherwise the number of carbons, oxygens and hydrogens are identical. The structural differences between the two sugars seem insignificant, but from the standpoint of practical organic chemistry, the conversion of galactose to glucose does not easily occur. However, the conversion can be accomplished biologically and this is demonstrated by most of us nearly every day if we ingest milk or substances containing galactose.

## *Normal digestion*

Digestion breaks down food into the different elements that the body requires for continued function. Foods are classified as proteins, fats and carbohydrates. Before the body can utilize them, it is necessary to change the food into molecules small enough to pass readily through the intestinal walls into the blood and lymph. After this process, the body uses the substances to build new tissues, to repair old and worn tissues, and to provide fuel for energy.

The agents which bring about digestion are enzymes. In the galactosemic patient we are interested in the carbohydrates. When they are digested, they are hydrolyzed to monosaccharides (simple sugars). The enzymes are specific in their action; that is, an enzyme will



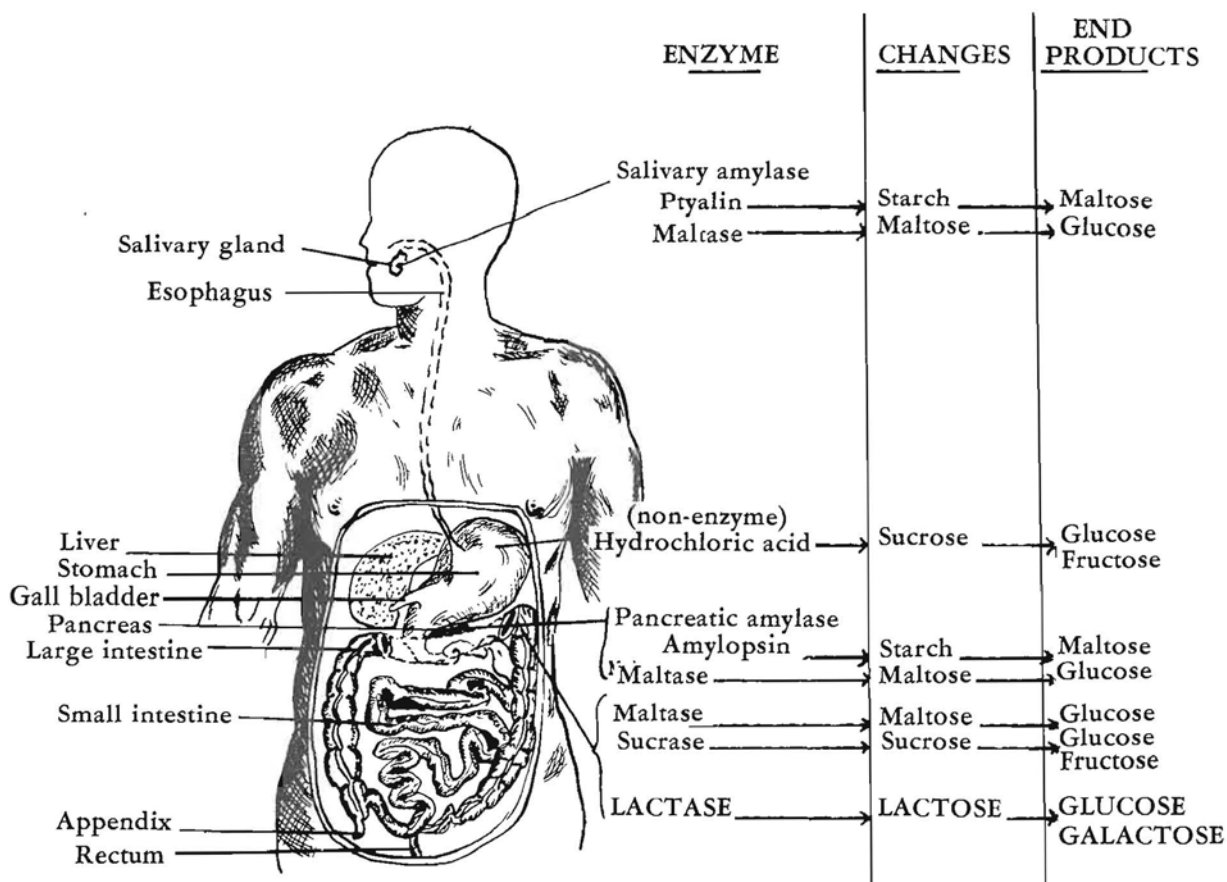


Figure 23.—DIGESTION OF CARBOHYDRATES ILLUSTRATING THE METABOLISM OF MILK LACTOSE IN GALACTOSEMIA.

"Nutrition in Health and Disease"; Cooper, Barber & Mitchell.

stimulate only one chemical reaction (see figure 23).

After food is swallowed, it passes down into the esophagus. Muscular contractions (peristalsis) force the food through the esophagus into the stomach. From the stomach, after partial digestion, it passes into the small intestine. Here digestion is completed. It is in the small intestine that the blood absorbs food substances and solid wastes enter the large intestine and are eliminated through the rectum. Liquid wastes are collected from the body cells by the blood and brought to the kidneys for excretion.

**Metabolism** is the chemical change taking place in the body after food is absorbed from the alimentary tract. It does not include

digestion. As noted in the diagram, carbohydrates are digested in the alimentary tract, forming the monosaccharides (glucose, fructose, and galactose). These monosaccharides pass through the intestinal walls into the blood stream. Blood carries the glucose to all parts of the body where it is either oxidized to give heat and energy or stored in the tissues in the form of glycogen. For a time after the ingestion of food, fructose and galactose may be present in the bloodstream but normally these are soon converted into glucose or glycogen. Glucose is always present in the blood and tissues. When the body is in a well-nourished state, glycogen is present in the liver and muscles. The liver has the ability to form glucose from sugar, from amino acids obtained

from proteins, and also to liberate glucose into the bloodstream during the time that food is being absorbed from the alimentary tract.

Dr. Herman Kalckar, mentioned earlier, was the first to demonstrate the pathway of galactose (figure 24). Briefly, this is what happens. Galactose is ingested as lactose which is broken down into a molecule of glucose and galactose and is absorbed separately. The galactose that is absorbed is converted to a phosphorylated sugar, just as glucose has to be before it can be utilized by the various cells in the body. The galactose 1-phosphate then is incorporated into a nucleotide, called UDP.

UDP, with the release of glucose 1-phosphate, contains a glucose molecule within its structure and an exchange is mediated by this enzyme system. This is the transferase system and the transfer occurs between galactose and glucose with a release of glucose which travels the usual pathways of metabolism for energy. Another important step is the fact that the galactose that is incorporated into this nucleotide can then be reconverted by another specific enzyme to the original UDP glucose which can then be reutilized. Dr. Kalckar demonstrated

that the defect in galactosemia resides at point (B) in figure 24. The phospho-galactose-uridylyl transferase, or the enzyme necessary for taking galactose and incorporating it into the nucleotide with release of glucose, is usually inactive in galactosemic individuals. As a result, metabolism stops and the metabolism of galactose 1-phosphate thus accumulates. Dr. Kalckar and his associates established that phospho-galactose-uridylyl transferase was inactive not only from the liver of the affected child, but in the red blood cells as well. Continued experiments have confirmed that the level of the enzyme in red blood cells is a reliable index to the clinical state. Now in use is the *transferase assay* or the *enzyme assay* which is suitable for quantitative measurement of levels of enzyme activity known to be characteristic for the galactosemic child, the carrier of the galactosemic gene, and the normal person as well.

Normal individuals have a value of approximately 5.9 to 6 units. The galactosemic child, for practical purposes, has no enzyme activity. A blood transfusion from a normal donor can transmit the enzyme activity of the transfused cell to a galactosemic child.

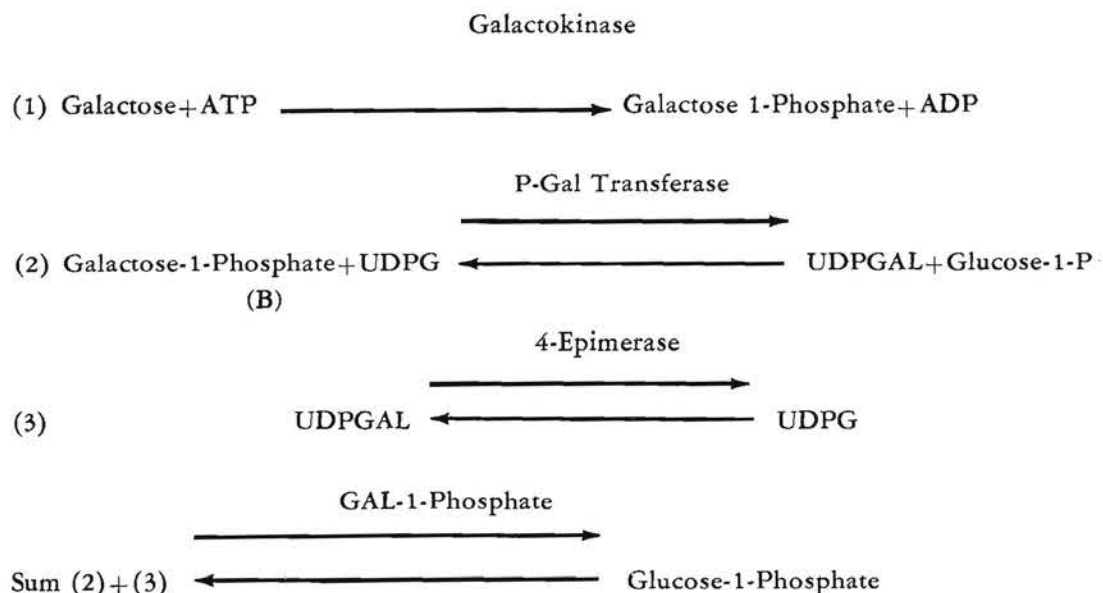


Figure 24.—METABOLISM OF GALACTOSE.

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It is not possible at the present time to prove that galactose-1-phosphate is the direct cause of the physiological effects observed. But various bits of evidence strongly suggest this assumption. Relatively high concentrations of the sugar are found in all tissues of the affected individual. In vitro experiments have shown that galactose-1-phosphate is inhibitory to a number of enzyme reactions in the body, particularly those related to glucose metabolism.

Certain tissues, such as liver, kidney, spleen, and brain appear to be more susceptible to damage than others. The earliest manifestations of liver damage are jaundice and liver enlargement (hepatomegaly). Cirrhosis of the liver slowly develops if the patient continues to ingest galactose. Such children show a typical picture of failure to thrive with lack of weight gain, poor growth, and feeding difficulties. They are very susceptible to infection, and death from overwhelming septicemia may occur in the first few days of life. In other children, death may occur from liver or kidney function failure or kidney malfunction. Other indications are protein in the urine and the presence of amino acid and sugar in the urine. This is indicative of damage to the absorptive mechanism of the renal tubule. In some cases the manifestations just noted are less severe. There may be unspecified failure to thrive or the finding of cataracts and mental retardation. Very few galactosemic patients are discovered in surveys of institutions for the retarded. This reflects the high mortality rate among the untreated children in their early years. Clinically the child may show several physical symptoms. He may appear to be well nourished. Actually the abdomen is swollen and distended due to an accumulation of fluid. The area around the eyes may appear puffy, the backs of the feet appear swollen and the liver is extremely large.

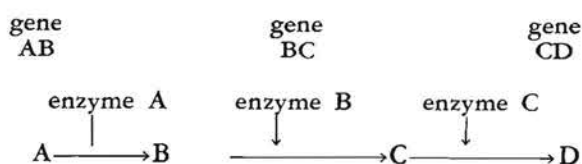
### Treatment and diet

Currently, galactosemia is treated by the very direct approach of removing galactose from the diet.

Tests have shown that the lack of dietary

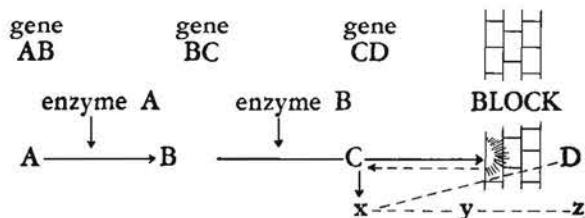
galactose in these patients has not been detrimental. Since such children have normal physical development, if treatment is begun early in life, abnormal neurological manifestations are absent and mental development appears normal. This seems to indicate that galactose from outside sources is not necessary for normal growth and development. The body seems to produce enough galactose from within for proper growth.

### NORMAL METABOLISM



Normally the sequence of reaction, A, B, C, and D, occurs uninterrupted in the body digestion to change or convert glucose into liver glycogen. A different enzyme regulates each stage.

### ABNORMAL METABOLISM



The stages from A, B, C, are normal, but then a block or defect occurs and the sequence cannot continue to D. When this happens, too little of the end product D is formed. C accumulates damaging the tissues and causing the formation of alternate pathways to form new substances—x, y, z.

In the galactosemic, if treatment is instituted before physical symptoms appear, the pattern of development compares favorably with normal children. If the diagnosis is delayed, the effectiveness of treatment depends upon how much damage has already been incurred. With treatment—*removal of lactose from the diet*—the kidney and liver manifestations disappear. Early eye cataracts may improve.

While it is clear that galactose-containing products must be avoided by the galactosemic, other galactose-containing sugars (oligosaccharides) are present in some dietary materials, such as soya beans, beets, and peas. Their effect on the galactosemic child is not known at this time. Lactose, as a readily available sugar, is used in drug products and it may be a constituent of food not suspected by the consumer. This makes the home management of a galactosemic child sometimes difficult.

It is now possible to actually measure erythrocyte galactose-1-phosphate and detect dietary breaks which would have been unsuspected otherwise. For example, one small galactosemic child showed evidence of an elevated galactose-1-phosphate level in the blood. Upon investigation it was discovered that she was, unknown to the parents, drinking the saucer of milk intended for the cat. Another child consumed some ice cream and cake while the parents were in the other room. Still another child was receiving milk at school.

Rapid technical advances have resulted in improvement in management of galactosemic individuals. The task now is to improve diagnostic procedures so that individuals with galactosemia can be detected early enough for treatment to be beneficial.

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# THE PREDICTABILITY OF GESELL DEVELOPMENTAL SCALES IN DOWN'S SYNDROME

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DR. ARNOLD L. GESELL, born in 1880 in Alma, Wisconsin, became famous as a pediatrician for his studies concerning the behavior of infants and children (1). He found that most children followed a somewhat similar pattern of development as they matured. The results of his work may be found in his *Atlas of Infant Behavior* which is now widely applied in child guidance and parental education.

The scales developed by Dr. Gesell and his associates at Yale University are probably the best known and most widely used instrument for the developmental evaluation of infants as young as 4 weeks of age. The developmental scales provide a means of evaluating the levels of behavior of the infant in four major areas over an extended period of time. These areas are:

1. Motor behavior.—evaluation of both gross and fine motor reactions, progress in sitting, walking, creeping, how he grasps for objects and handles or manipulates them.
2. Adaptive behavior.—evaluation of how the infant reacts in certain circumstances—visual-motor reactions and the solutions of problems, e.g., how does the infant react to the ringing of a bell or an object dangled in front of him?
3. Language behavior.—concerned with all means of communication including reactions to attempts at communication by others, changes in facial expression, posture, gestures.
4. Personal-social behavior.—involves the infant's reactions to the particular social en-

vironment in which he is reared, his social manners, response to others, progress in "self-help" activities.

No single score is computed for the test. The examiner presents test objects to the child and evaluates the responses made. Each of the above four developmental areas is evaluated, and a *developmental age* in months is assigned to the infant's performance in each. This is done by comparing, on a subjective basis, the infant's reactions with the norms for infants and children at ages 4, 18, 28, and 40 weeks; and at the ages of 12, 18, 24, and 36 months (2). According to Gesell's developmental levels a normal infant will show the following patterns (3, 4):

## Months

- 1----- Begins to have some regard for his surroundings. Can raise his head.
- 2----- Begins to smile and vocalizes by cooing.
- 3----- Has some head control and turns head in direction of sound.
- 4----- Grasps objects with both hands.
- 5----- Rolls over.
- 6----- Begins to transfer objects from hand to hand.
- 7----- Sits alone.
- 8----- Creeps.
- 9----- Pulls up and begins to use forefinger and thumb apposition.
- 10----- Stands holding on.
- 11----- Begins to walk holding on.
- 12----- Stands alone and begins to use single words.



### Months

- 12-15---- Walks alone and creeps upstairs.  
15-18---- Walks and rarely falls; begins to throw a ball.  
18-24---- Runs, climbs, jumps, walks upstairs, and can kick a ball. Points to various parts of his body; can use two words together.  
24-36---- Builds a tower of three or more blocks, can be taught to put away toys in proper place. Folds paper once, imitatively, listens to stories illustrated with pictures. Asks for things at table by name, opens doors, and helps to undress himself.  
36-48---- Repeats two numbers and enumerates the objects in a picture. Knows own name and repeats a sentence of six syllables. May try to sing, run, jump, ride a tricycle, and dance. Attempts to draw pictures and string beads. Plays simple games. Can unbutton clothes, wash hands, and sometimes brush teeth. Does not wet bed as a general rule.

Gesell divides the life span of an individual into six periods of development (4).

1. Fetal and embryonic--- Conception to birth
2. Neonatal ----- Birth to 2 weeks
3. Infancy----- 2 weeks to 2 years
4. Childhood:
  - Early preschool----- 2 to 6 years
  - Late school years---- 6 to 12 years
5. Adolescence----- 12 to 18 years
6. Adult ----- Over 18 years

Of course, deviation from these norms may not necessarily be considered abnormal. Some infants are simply slower than others in their development, but usually catch up and are considered perfectly normal in their physical and mental functioning (3).

It is noted that the Gesell scales rely heavily on the motor and physical functions of the child. It does not evaluate intellectual

capacity. Therefore, in the first year of life it is not a good predictor of a child's later mental potential. Using the developmental age assigned and the child's chronological age, a *developmental quotient* is determined.

$$\frac{MA}{CA} \times 100 = DQ$$

Maturation age divided by chronological age, multiplied by 100 gives the developmental quotient.

Caution must thus be exercised in interpreting the developmental quotient (DQ). They are not the same as intelligence quotient (IQ) scores. However DQ scores at the age level 2½ to 4 years tend to correspond rather closely with IQ estimates (3).

There is much controversy concerning the usefulness of Gesell developmental scales (5). Some investigators have criticized the predictive value of Gesell's developmental scales and some the usefulness of developmental scales of any type during infancy (8, 10). Knobloch (11) has emphasized that their greatest value is when they are used as a neurologic tool, and that they are not to be used to predict later intellectual capacity as measured by intelligence quotients. Illingworth (12) has also emphasized their usefulness in establishing the presence of mental retardation in early childhood.

### Study of mongolism

Over a 7-year period the Gesell examinations have been used in the Child Development Clinic at Los Angeles Children's Hospital for testing and retesting 76 infants diagnosed as having Down's Syndrome (mongolism). Initially, observations of these infants raised doubts as to the validity of predictions made on the basis of the first year's developmental scales (7) and also raised the question as to whether progressive retardation occurs in Down's Syndrome (6). A preliminary investigation by Share, Webb, and Koch (7) suggested that developmental quotients obtained during the first year of life have little, if any, predictive

value. Developmental quotients and maturity age scores obtained during the second year of life appeared to be valid predictors of future development up to 3 years of age. The study also suggested the development of Down's Syndrome infants follows a pattern of slow, steady improvement, but the initial sample size was too small for the data to be completely reliable (5).

The second study (5) using 76 infants extended the longitudinal observations into the fourth year of life. Of the initial 76 infants, only 53 were suitable for study. Age of less than 1 year and the willingness of the parents to participate in the study were the only criteria of acceptance for study. A Gesell examination was performed at least once each year over a period of 3 years. Eight subsequently died, 8 moved, and 6 were lost to the study which left 31 subjects who were tested regularly over a 3-year period.

Twenty-three children were followed from infancy to 48 months of age. For statistical purposes, the children were tested during four age groups:

Group I-----	2 to 13 months
Group II-----	14 to 24 months
Group III-----	26 to 36 months
Group IV-----	38 to 48 months

All lived at home except for three institutionalized children.

## Results

In the sample of 31 children, both maturity age scores and developmental quotients at all of the first three age levels showed significant differences. As in the earlier study, the DQ scores reflected a regression, and the MA indicated slow but steady growth. No developmental plateaus were evident in the analysis of the MA scores, i.e., the MA scores did not show arrested maturation.

With a smaller sample of 23, but with an extension of the investigation into the fourth age group (36 to 48 months), the MA scores continued to be significant in the direction of

slow, steady growth. However, it was noted that there was a progressive deceleration in the rate of maturation as measured by MA scores. The difference between Groups I and II shows a mean maturity age increase of 6.26 months. Between Groups II and III, the amount of increase diminishes to a mean score of 4.87. The amount of increase between Groups II and IV decreased to 3.57. This deceleration is at the rate of approximately 1.35 months per year.

When one inspects the DQ data in the chart (figure 25), there does appear to be progressive retardation which may mean to some a deterioration or an arrested development.

However, when one looks closely at the maturity age in the chart in figure 26 it is quite apparent that steady developmental progress occurs.

The rate of progress, however, decelerates significantly. During the first 12 months, the average infant gained 6.26 months as compared to only 4.87 months during the second year and 3.57 months during the third year. This rate of deceleration early in childhood is a curious phenomenon. It is possible that this observation supports the vague hypothesis that children with Down's Syndrome age prematurely or that some metabolic event is suppressing their normal rate of maturation. These children usually do not test abnormally low during the first year of their life, (3) but by the time they reach the third year their mental achievements drop considerably and most are moderately retarded. This initial progress probably explains why many parents who have a child without obvious stigmata are reluctant to accept a diagnosis of mongolism and prefer to shop around from one clinic to another, in the hope that the diagnosis is wrong. While many mongoloid children reach a mental status ranging from 3 to 7 years, the rate of their progress is significantly slower than that of normal children. Thus, it is not unusual to see some of these children who, at 3 years of age, are barely beginning to walk or still have no speech at 4 or even 5 years. However, most mongoloids eventually learn to walk and talk, even though their speech may be significantly limited. Rarely are these children able to go into the regular classes of the public school

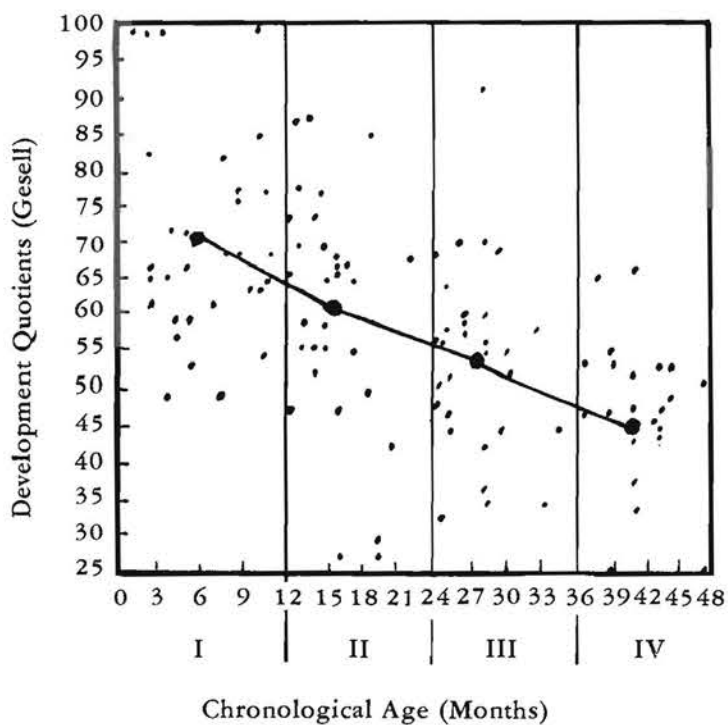


Figure 25.—SCATTERGRAM OF DEVELOPMENTAL QUOTIENTS OF MONGOLOID SAMPLE. GROUPS I-III, NO. —31; GROUPS I-IV, NO. —23.

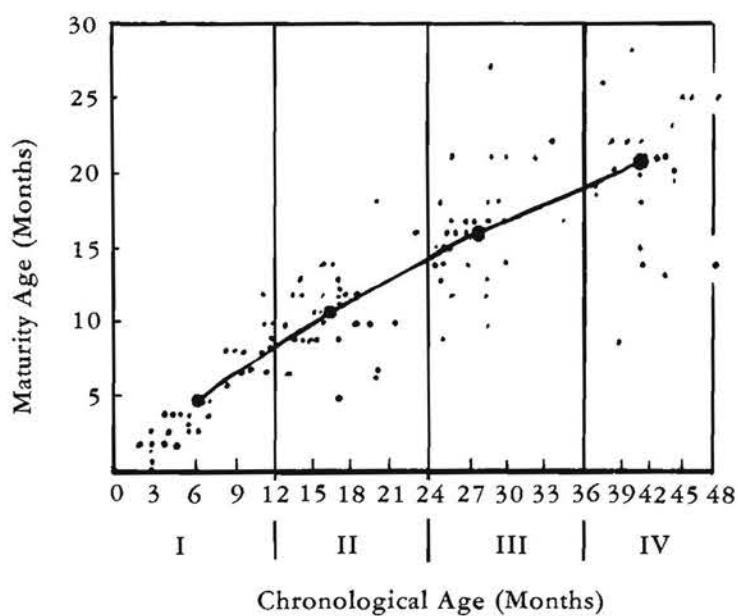


Figure 26.—SCATTERGRAM OF M.A. IN INFANTS WITH MONGOLISM (DOWN'S SYNDROME). GROUPS I-III, NO. —31; GROUPS I-IV, NO. —23.

system, although a good many are able to qualify for enrollment in the special classes designed for trainable and educable mentally retarded children (3).

Much more data (5) is needed before the full meaning of these findings can be clarified. The data thus far does not indicate when a plateau in the mental age of such children occurs. Gibson (13) suggests that the IQ is relatively stable within the age range of 5 to 19 years. If so, perhaps a true plateau of development does not indeed exist.

When additional data are obtained by correlating developmental and intelligence quotients obtained by Stanford-Binet testing, the full value of developmental testing in children with Down's Syndrome can be adequately documented.

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