Hearing in Children

Sixth Edition

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Foreword

Pediatric audiology came quietly into being in the early 1940s, spurred by dedicated educators of the hard-of-hearing and deaf who studied auditory behaviors of normal-hearing children and applied those comparisons to their students with hearing loss. These educators were Lord and Lady Ewing in England who developed the earliest behavioral techniques for testing the hearing of very young children. Today many thousands of individuals with hearing loss are enjoying more successful lives as a result of having been identified early in life.

Today we look back to this period of time when the potential of early infant development was finally recognized. Out of an era of mechanistic behaviorism advocated by B.F. Skinner, there gradually grew an understanding of the latent and hidden capabilities of the newborn human infant—capabilities that needed to be stimulated and nurtured in certain ways at certain times to reach full maturity. Foremost in fostering this understanding were individuals such as Chomsky, Piaget, Lennenberg, Apgar, Spock, and Brazelton, and their many, many students. The thoughts, works, and contributions of these clinicians/scientists have extended the purview of the developing infant to other related professionals while recognizing the important roles played by the parent, caregiver, and family.

Nearly all health and education professions have a stake somehow in the human newborn infant; hardly a specialty exists that does not claim a part of this emergent individual. Pediatric audiologists may focus their attentions on the hearing mechanism of the infant, but hearing does not exist in a vacuum; it is part of a complicated, interrelated, living, breathing, thinking child that sustains the auditory response and, in turn, is modified by it. The skills and knowledge that are most interrelated with hearing are linguistics and speech. Both develop in the normal infant through the passive act of listening, albeit in different ways. Normal development and production of speech is dependent on the presence of normal hearing. Most children with normal hearing develop appropriate speech production skills during their early childhood years. For children with hearing loss, speech production intelligibility is directly proportional to the degree of the loss. In other words, the better the hearing, the better is the child's speech.

This is not so for language. Language skills of children with hearing loss appear to be fairly equally affected by almost any degree of significant hearing loss. We have now confirmed that the only factor that significantly affects language abilities in young children with hearing loss and deafness is the time of intervention. In other words, once again, we see that the earlier the intervention, the better the child's language skills.

All children with hearing loss, even those with mild and moderate hearing losses, will show delay in receptive and expressive language. For this reason, all children with hearing loss must be identified early enough in the earliest months of life to enable successful early intervention. The highly technical advancements in hearing aids and cochlear implants provide access to the auditory world even for babies with profound sensory deafness. Early identification followed by early interventions takes advantage of these infants' young brains to help them become fully functional, hearing individuals.

Many thousands of individuals with hearing loss are enjoying more successful lives as

a result of having been identified early in life by audiologists. Make no mistake: our goal is for all! Not just the child with parents who can afford testing and monitoring. Not just those children who happen to fall into a category that places them at risk for hearing loss or profound deafness. Not just the infants that happen to be in the right place at the right time to be screened and evaluated. We are talking about all children with hearing loss. They are all entitled to be touched by the hands of pediatric audiologists using the best in modern technology to accurately and promptly diagnosis and manage these babies with hearing loss. There is too much at stake to miss any of these youngsters.

The field of medicine gives us an interesting analog: it has been said that "medicine is unique among the sciences in that it strives incessantly to defeat the object of its own invention." This object, of course, is disease, and there is a parallel in audiology. Since our beginnings in the mid-1940s, we have measured, described, researched, cataloged, analyzed, and synthesized the entity of hearing loss exhaustively. Now, having defined it completely, we must busy ourselves with preventing the devastation of its effects on children. Such prevention can only be accomplished by early detection of the condition and by proper provision of early intervention, appropriate therapy, and purposeful education. Armed with our new depth of knowledge, we can assuredly enhance and enrich the lives of children with hearing loss and their families.

These observations challenge us to utilize the particular skills that we acquire during our education and training. Our unique knowledge of the interrelated aspects of hearing, speech, and language is an important contribution to understanding the wonder of communication development in infants. The audiologist's goal is to identify hearing problems and to understand the accompanying auditory disorders—an especially challenging task in infants and young children. To this end, we must constantly hone our skills and technologies. Our ultimate goal is that all infants and young children with hearing loss will achieve their full potentials and attain the best future possible as happy, successful, and productive adults.

—Marion P. Downs, DHS, DS
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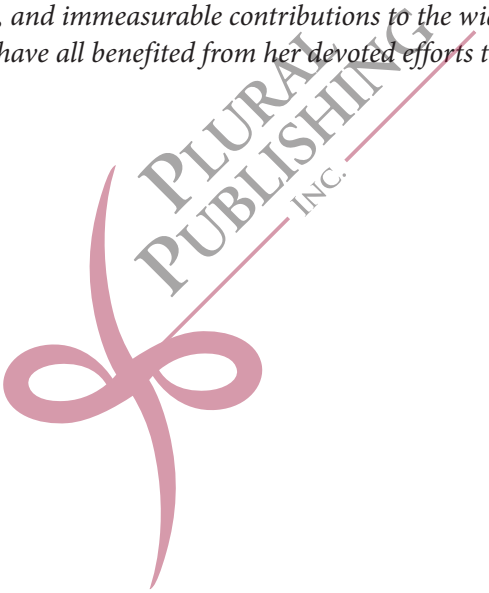
This final edition of Hearing in Children *is dedicated to the legendary Dr. Marion P. Downs who contributed more than 60 years of her career to bring the world of pediatric audiology from its very basic beginnings to today's unlimited promise for all children with hearing loss. As my office colleague and close friend throughout those many years, she continues to inspire us as she serves as our role model even as she reaches her centennial year of life. Marion Downs has changed the world for countless children, families, and professionals; we are grateful for her wisdom, guidance, and immeasurable contributions to the wide arena of pediatric audiology. We have all benefited from her devoted efforts to better our lives.*



CHAPTER 2

Early Development

There are few events in life that surpass the birth of a new baby. The mystery and beauty of the origins of life have been well documented in books, videos, and the Internet, and yet, with each new birth, we continue to be awestruck. The time leading up to the normal birthing process is generally 266 days (38 weeks) from conception to birth, although it should be noted that only about 5% of births occur on the actual due date. Parents typically develop growing anticipation as the delivery time draws near. And, the satisfaction of taking the new baby home is an emotion that remains strong throughout life. Birthing is a miraculous event that produces an amazing 97% of normal births in spite of the complexities of prenatal development.

However, the fact is that about 3%, or one in every 33 babies born in the United States, is born with a birth defect (NICHD, 2012). Prematurity and low birth weights are identified as the primary causes of birth defects. Many different factors may be associated with the development of birth defects, such as genetic and chromosomal aberrations, *in utero* exposure to viruses or bacteria, uncontrolled maternal diabetes, maternal cigarette smoke, maternal use of drugs and alcohol during pregnancy, and prenatal exposure to chemicals. All of these factors may influence normal infant growth or development, resulting in different types of birth defects. From the audiologist's point of view, each of these factors can result in an infant being born with hearing impairment as a single isolated defect or an associated symptom to other birth defects. It is important that audiologists have a full understanding of the numerous processes involved in the earliest embryonic and fetal development of organ systems in the newborn.

The embryologic development of the ear is of more than academic interest to the clinician. An understanding of embryologic relationships helps confirm a diagnosis and suggests the need for early hearing assessments. If one is aware of the timetable of prenatal development and the association of the various organs and structures with each other, the suspicion of deafness and its subsequent diagnosis and treatment become easier. The origination and major changes in the development of the ear and the hearing system take place in the mother's womb as the baby becomes a progressively more complex structure over time. Several processes occur concurrently to produce the final structure, including enlargements, constrictions, and foldings, which are further modified by evaginations and invaginations. However, development of the auditory structure does not cease, and is not totally complete, at the time of birth.

Knowledge of the origins of auditory structures (known as *phylogeny*) can be of diagnostic significance to the clinician. For example, when an infant presents with a congenital skin disorder, the clinician considers the fact that the skin and the otocyst (the primitive cochlear structure) both originate from ectoderm. It may then be logical to suspect that anomalies of the cochlear structures may have occurred contiguously with the skin disorder and that a search for severe sensorineural deafness is

in order. Similarly, the timing of development of the various organ systems guides the clinician to suspect that a hearing loss may have occurred at the same time that other systems were affected. A noxious influence on the fetus at 2 months of gestation may result in a malformation of the pinna developing at that time. The pinna malformation, however, does not necessarily imply malformation of the ossicles of the middle ear. Although, as you will see later in this chapter, the ossicles of the middle ear partially share the same time clock as the pinna in embryologic development, the basic origins of the structures are different; on the other hand, an insult to one may well result in a related insult to the other.

Principles such as these allow clinicians to look for the occult symptom of hearing loss whenever an overt embryologic-related symptom becomes evident. The prognosis for auditory function can then be evaluated with consideration of what is known regarding the origin of the structures in question and the expected auditory pathology (Jones, 2011).

BASIC PRINCIPLES OF GENETIC INHERITANCE

Humans take great pride in identifying distinguishing traits from one generation to the next. We enjoy speculating on the resemblance of children to their parents and grandparents. With such observations begins the appreciation of genetics. Genetics and the study of hereditary disorders are among the most rapidly evolving disciplines among the medical sciences. There is probably some genetic component in almost all disease conditions, but the extent of the genetic component likely varies. To provide competent services to infants and their families, audiologists need an elementary understanding of the basic genetics, cytogenetics and molecular genetics, the inheritance of hereditary disorders, specific disorders associated with genetic disease, and the process of genetic evaluations. Tremendous strides have been made in understanding the genetics of deafness during the past two decades. In 2002, about 200 syndromes associated with deafness had been typed with genetic origins; by 2006, investigators had identified the genetic identity of about 300 syndromes; in 2012 at least 400 genetic-based syndromes were described that are likely to include permanent hearing loss. It should be obvious that the study of clinical genetics is important as it allows investigators and clinicians to understand genetic transmission and to ultimately prevent and treat inherited conditions in the future.

It is important to distinguish among the many terms used in describing pediatric hearing loss. *Congenital* means present at birth; "congenital" does not make any implications regarding the etiology of the condition. Infants born with hearing loss, regardless of the cause, have congenital hearing loss. *Heredity* means inherited, or "passed down" from previous generations through DNA, genes, or chromosomes; *hereditary* implies chromosome or gene control of the condition in question. *Genetic* means caused by a gene; *genetic* may also be considered a special subset of hereditary. *Familial* means the symptom or sign is present in several related family members; however, the term *familial* does not in any way imply etiology.

Although hereditary disease can become apparent at any point in an individual's life span, genetic and chromosomal abnormalities and associated congenital malformations are especially important in pediatric specialties. The relative contribution of these disorders to infant mortality rate has increased as the prevalence of infectious childhood disease has decreased. In the pre-antibiotic era, most infant mortality was attributable to infectious disease; today, in

developed countries, most infant mortality is attributable to genetic disorders and congenital malformations. Of recognized pregnancies that end in spontaneous abortion, 50% to 60% have detectable chromosomal anomalies. It is estimated that 2% of all newborns demonstrate chromosomal or single-gene disorders. These abnormalities produce significant neonatal mortality and morbidity. Infants with genetic anomalies who survive into childhood require substantially greater health and educational care than nonaffected children.

DNA

Cells are the fundamental working units of every living system. All the instructions needed to direct their activities are contained within the chemical DNA (deoxyribonucleic acid). DNA is the base unit of heredity, "the code of life," or the molecular "letters" of inheritance. DNA from all organisms is made up of the same chemical and physical components. The DNA sequence is the particular side-by-side arrangement of bases along the DNA strand. This order spells out the exact instructions required to create a particular organism with its own unique traits. When changes or errors occur in this ordered sequence of the genetic code, disease may result. DNA is a nucleic acid consisting of four nitrogen-containing bases attached to a sugar-phosphate polymer and arranged on intertwining strands, the well-known "double-helix." The four bases, adenine (A), guanine (G), cytosine (C), and thymine (T), occur in predictable pairs on complementary strands such that A on one strand always pairs with T on its complementary strand, and G on one strand always pairs with C on its complementary strand. One set of complementary bases, <AT> and <CG> is a base pair. There are three billion base pairs of DNA that tell the body how to grow and build and how to repair itself. Because bases occur in these invariant pairs, one strand of DNA contains all the information necessary to construct its complementary pair.

DNA stores and encodes a vast amount of information based on the sequence of base pairs. For segment N bases long, there are N^4 possible base-pair sequences. The base-pair arrangement of DNA encodes the complete genetic information of an organism; it is called the organism's genome. Genomes vary widely in size: the smallest known genome for a free-living organism (a bacterium) contains about 600,000 DNA base pairs, while human and mouse genomes have some three billion.

Accurate replication and transmission of the genetic code are ensured by the base-pair coding and double-helix structure of DNA. Figure 2–1 shows the double-helix structure of DNA (upper half of figure) and replication (lower half of figure). During replication, the double helix unwinds and separates into two single strands with their associated bases. Each strand serves as the template for a new, complementary strand. For example, the unwound DNA strand at the bottom right of Figure 2–1 with bases <ATCACT> will direct synthesis of a strand with complementary bases <TAGTGA>. After replication, two "daughter" double helixes will result; each daughter will contain one original parent strand and one newly synthesized complementary strand. In addition, if a base on one strand is lost or damaged, it can be replaced using the complementary strand to direct its repair.

Genes

Genes are the fundamental physical and functional units of heredity. Genes are sequences of DNA that direct protein synthesis. A gene may contain several hundreds or even thousands of base pairs of DNA.

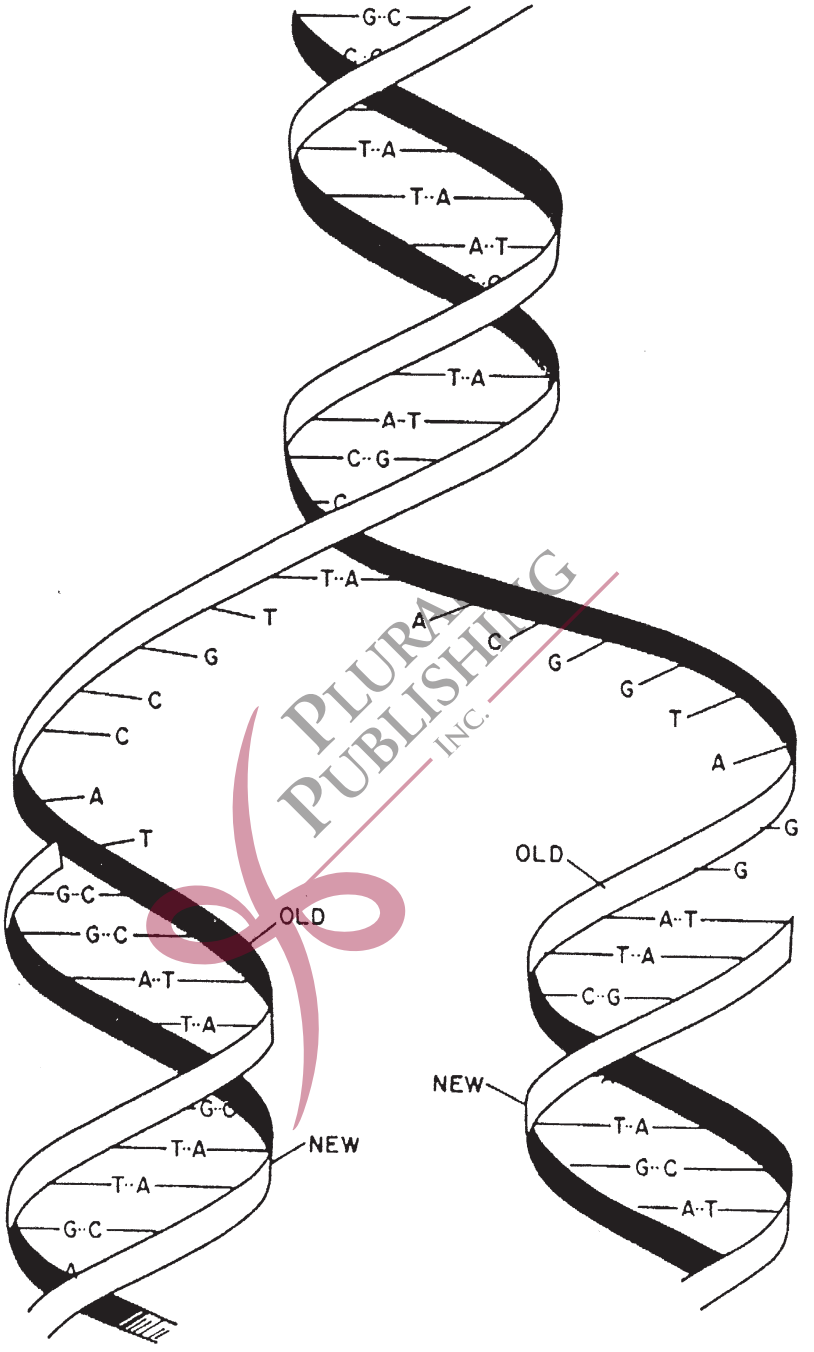


Figure 2–1. DNA, showing base-pair arrangement on intertwining sugar phosphate strands creating the double-helix structure of DNA. As the double helix unwinds and separates into single strings, each strand serves as a template for reproducing the complementary strand. With permission from *Clinical genetics and genetic counseling,* 2nd ed., by T. E. Kelly, 1986, Chicago, IL: Year Book Medical.

Genes are the main working parts of DNA; they instruct the body to perform specific functions. A mutation in even a single base pair may result in inaccurate synthesis of an essential protein and lead to genetic disease. Approximately 60% of hearing loss is estimated to be due to mutations in genes. This can be further broken down into syndromic hearing loss, estimated at about 30% of genetic hearing loss, and nonsyndromic hearing loss comprises the remaining 70% of hereditary hearing loss (Avraham, 2011).

Humans have about 20,000 genes. Genes are ordered in a linear fashion on uniquely identifiable structures known as chromosomes, contained in the nucleus of all cells. Genes, by themselves, do not directly make us who we are. Instead, the genes produce proteins that are dispatched throughout the body to execute the genetic will; if the genes are the blueprints, the proteins are the working parts (*Time,* May 20, 2013). Because genes occur at a specific locus on a chromosome, in many cases it is possible to identify the precise gene responsible for a specific genetic disorder (Keats, 1996). For example, researchers have determined that individuals with neurofibromatosis type II (bilateral acoustic neuromas) demonstrate an abnormal gene on chromosome 22; individuals with Waardenburg's syndrome demonstrate an abnormal sequence of DNA on chromosome 2; and individuals with Huntington's disease have an abnormal prolongation or repetition of a particular DNA sequence on the tip of chromosome 4.

Chromosomes

Chromosomes are long segments of DNA containing hundreds of genes. They are observed by light microscopy in nucleated cells undergoing active cell division. Chromosomes are categorized by their distinctive sizes and shapes. Prior to cell division, the chromatids separate, and one chromatid contributes a complete copy of DNA to each daughter cell. Chromosomes manifest this appearance during only one phase of active cell division. During other phases of the cell cycle (growth and protein synthesis), chromosomes are uncondensed, single, elongated strands. Figure 2–2 is a diagrammatic representation of a chromosome during one phase of cell division. At this stage, the DNA has condensed and doubled; the chromosome appears as two longitudinal rods, called chromatids, joined at a single point called the centromere. The chromatids each contain one complete copy of the

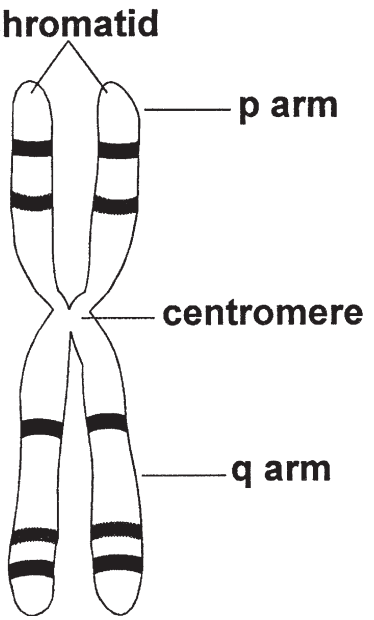


Figure 2–2. A chromosome during cell division. DNA has condensed and doubled. The two longitudinal halves of the chromosome are called chromatids, each of which contains a complete copy of DNA. The chromatids are joined at the centromere that divides the chromatids into unequal arms, the short (p) arm and the longer (q) arm.