Numerous chromosomal and genetic disorders exist, and several volumes would be needed to delineate each of them. For the purposes of this chapter, the authors have chosen to focus largely on several chromosomal disorders, mainly the trisomies. Other common genetic disorders will be briefly referenced and sources will be provided for those interested in understanding these disorders in greater detail.

About 1 in 150 babies is born with a chromosomal abnormality (American College of Obstetricians and Gynecologists, 2001, 2005; Carey, 2003). These disorders are caused by errors in the number or structure of chromosomes, which usually result from an error that occurred when an egg or sperm cell was developing. Babies may be born with too few or too many chromosomes. In some instances, a piece of a chromosome may be missing or the chromosomes may be rearranged. However, it is still unknown why these errors occur. Nonetheless, these errors can cause a variety of birth defects ranging from mild to severe.

Many children with a chromosomal abnormality have mental and/or physical birth defects, ranging from mild to severe. In addition, some chromosomal abnormalities result in miscarriage or stillbirth. Research indicates that nothing that a parent does or does not do during pregnancy can cause a chromosomal abnormality.

A common type of chromosomal abnormality is a trisomy, where there are three copies, instead of two, of a specific chromosome. In most instances, an embryo with the wrong number of chromosomes does not survive and a miscarriage results, often during the first trimester. In fact, 75% of first trimester miscarriages are caused by chromosomal abnormalities in the embryo (American College of Obstetricians and Gynecologists, 2005).

Other errors can also occur, usually prior to fertilization. Alteration in the structure of one or more chromosomes can result. Chromosomes may be deleted, misplaced, inverted, duplicated, or exchanged with part of another chromosome. These structural rearrangements have no effect at times if all of the chromosome is present, but is just rearranged. However, an increased risk of miscarriage or birth defects exists nevertheless.

Errors in cell division may also occur after fertilization, resulting in mosaicism, a condition in which an individual has cells with different genetic makeups. The severity of the condition is dependent primarily on the percentage of abnormal cells. As result, some people are only mildly affected while others are profoundly impacted.
Down syndrome (DS) is one of the most common chromosomal abnormalities, affecting approximately 1 in 800 babies (American College of Obstetricians and Gynecologists, 2005; National Down Syndrome Society, 2006). Significantly, the risk of DS and other trisomies increases with the mother’s age. According to the American College of Obstetricians and Gynecologists (2005), a woman’s risk of having a baby with DS is 1 in 1,000 at age 30, 1 in 400 at age 35, and 1 in 100 at age 40.

In addition to mother’s age, other parental characteristics may be associated with an increased risk of certain congenital malformations in offspring. Research suggests that parental occupation and Hispanic ethnicity may increase the risk of some birth defects. A case–control investigation of babies born in Texas from 1996 through 2000 revealed that maternal occupation as a cook or nurse was related to an increased risk of oral clefts and neural tube defects among births to Hispanic women, but not among births to non-Hispanic White mothers (Brender et al., 2008). Hispanic fathers who were electricians had a greater risk of having babies with chromosomal abnormalities, particularly trisomy 18. These associations did not occur among the babies of non-Hispanic White fathers.

Maternal smoking and other behaviors may increase the risk of chromosomal abnormalities. For example, in their investigation of the effects of maternal smoking on prenatal screening for DS and trisomy 18 in the first trimester of pregnancy, Miron et al. (2008) demonstrated that maternal smoking was associated with an increase in average risk and rates of trisomy 18, but not an increase in the average risk and rates of DS.

Babies can also be born with an extra chromosome 13 or 18, both trisomies which are significantly more severe than DS. Carey (2003) reported that about 1 in 10,000 babies are born with trisomy 13 (Patau syndrome). SOFT (2006) reported that 1 in 6,000 babies are born with trisomy 18 (Edwards syndrome).

As a result of new technology, some uncommon chromosomal abnormalities have been identified. In some of these cases, the abnormalities are so rare that only a few children may be impacted. Some of the uncommon abnormalities include deletion, in which a small section of a chromosome is missing and microdeletion, in which a single gene or minute amount of material is missing. In addition, a translocation occurs, in which a section of a chromosome is attached to another chromosome, or an inversion takes place, in which a section of a chromosome is inserted upside down. Furthermore, duplication occurs, resulting in extra genetic material, or a ring chromosome develops, where material is deleted at each end of a chromosome and the new ends then join to form a ring.

Some of these abnormalities, particularly inversions and translocations, may not impact a child’s health if no genetic material is missing or duplicated. However, deletions, even small deletions, often result in severe complications. Prader–Willi syndrome, Cri-du-chat syndrome, Wolf–Hirschhorn syndrome, and 22q11 deletion syndrome are well-known examples.

Typically, the risk of parents having another child with a chromosomal abnormality is low. Carey reported that parents with a child with DS had a 1% risk of having another DS baby, if the mother is younger than 35 years of age. Those
mothers over 35 have a risk similar to others of their age. Nonetheless, there are exceptions.

Early diagnosis and intervention are essential. Genetic counseling is often recommended and supportive counseling for the parents is usually helpful, particularly group support. In some cases, early education intervention, particularly with non-fatal disorders like DS or Klinefelter syndrome, is vital. Furthermore, expert medical care is necessary.

Down Syndrome (DS)

Down syndrome (DS) is a chromosomal condition related to chromosome 21. The extra number 21 chromosome interrupts the normal course of development, causing the characteristic clinical features. However, some individuals have an extra chromosome 21 in only some of their cells, resulting in mosaic Down syndrome (MDS). In addition, a small number of people with DS have the syndrome because part of chromosome 21 translocated to another chromosome, either before or at the time of conception. As a result, these individuals have two copies of chromosome 21 and additional material from chromosome 21 attached to another chromosome.

DS is typically not inherited. MDS is also not inherited, but is the result of random error during cell division, resulting in some cells having an extra copy of the chromosome. Translocation DS may be inherited.

DS affects 1 in 800 to 1 in 1,000 live born infants (National Human Genome Research Institute, 2008). Individuals with DS have learning problems, mental retardation, poor muscle tone in infancy, as well as a characteristic facial feature. They also have an increased risk for having heart defects. Approximately 40–60% of those born with DS have a heart defect (Drew, 1990).

Those with DS also have more digestive problems, including gastroesophageal reflux or celiac disease. Some infants with DS have problems with swallowing or they can experience bowel blockage. Some also have hypothyroidism.

DS children often have frequent colds as well as sinus and ear infections. Others experience sucking problems during infancy, as a result of poor muscle tone. Sucking problems create difficulty with breast feeding initially. Furthermore, some infants have eye problems, most notably strabismus and cataracts.

Intellectually, DS children may function from a low-normal to a mentally handicapped range. Wisniewski et al. (1996) found varying degrees of cognitive and other neurological dysfunction in DS children. Furthermore, Anderson (1998) and Nadel (1996) have also commented extensively on the wide range of intellectual functioning.

Researchers have evaluated language comprehension in children with DS and other disorders. Price et al. (2007) compared the receptive vocabulary, grammatical morphology, and syntax skills of DS boys with those of boys with fragile X syndrome (FXS) (who were also classified as having autism, autism spectrum disorder, or no autism) and typically developing (TD) boys. The DS boys performed lower in language comprehension than the FXS boys without autism and TD boys. DS
boys and FXS boys varied in receptive language performance, indicating unique language patterns for each disorder. The investigators recommend targeting language comprehension in assessing and treating these groups.

Ferrier et al. (1991) investigated the conversational skills of persons with FXS, compared to two matched groups of individuals with autism and DS. The researchers found that the FXS group relied on more eliciting forms in conversation than the DS group. The FXS group also used partial self-repetition more frequently than both the DS and autism groups. The DS group exhibits more speech dysfluencies than the autism group, but no more than the FXS group. Compared to the other groups, the autism group used more inappropriate phrases.

Research on the phonological accuracy and speech intelligibility has shown differences based on the type of disorder. Based on a sample of boys with FXS with autism spectrum disorder (ASD), FXS only, DS, and typically developing (TD) boys, Barnes et al. (2009) discovered that DS boys had lower scores on phonological accuracy and the occurrence of phonological processes than all other groups of boys. DS boys also relied on fewer intelligible words than did boys in the TD group. The investigators concluded that DS boys exhibited more extensive delays in all phonological indicators than boys in the FXS and TD groups.

Wright, Lewis, and Collis (2006) found that object search and play behaviors of children with DS rely more heavily on imitation than is the case for typically developing children. In another investigation, Landry et al. (1994) related that DS children initiated fewer exchanges in less structured situations than normal peers, but were more likely to increase compliance with directive requests.

Cicchetti (1991) expressed concern about the biologically based risk factors on early self-development, particularly self-concept, in children with DS. Dykens et al. (2007) examined the self-perceptions, thoughts, hopes, and inner lives of children with DS and Prader–Willi syndrome (PWS). Positive and negative self-appraisal was found to be related to syndrome status or maladaptive behavior. Hodapp et al. (1997) reported that the amount of the child’s maladaptive behavior is highly correlated with parental stress and that the parents of DS children generally report lower levels of stress, related to the degree to which the child is reinforcing and acceptable to the child. Finally, Ghosh et al. (2008) indicated that ASD manifests as a distinct behavioral phenomenon in DS, and DS children with ASD display more restricted repetitive and stereotyped patterns of behavior, interests, and activities.

**Prader–Willi Syndrome (PWS)**

First reported in 1956, the Prader–Willi syndrome (PWS) occurs when there is a paternal deletion on chromosome 15, a maternal uniparental disomy, or an imprinting defect. According to the Prader–Willi Association (2008), every case of PWS is the result of the baby failing to receive active genes from a specific section of the father’s chromosome 15. Approximately 70% of all cases are the result of paternal deletion or microdeletion. Usually a deletion occurs for no known reason and there
is less than a 1% chance of reoccurrence. About 25% of all cases are the result of maternal uniparental trisomy, where the developing embryo begins with three copies of chromosome 15 and one of the copies is “lost,” the chromosome 15 from the father. The result is the same as deletion, even though there are two complete copies of the mother’s chromosome 15. Finally, less than 5% of all cases results from an imprinting defect. In these instances, the PWS genes on the father’s chromosome are present but do not work because the imprinting process is faulty.

PWS occurs equally in both genders and all races and has a prevalence of 1 in 12,000–15,000. The major characteristics include hypotonia, hypogonadism, hyperphagia, cognitive impairment, and problematic behaviors. The major medical concern is morbid obesity, due to a tendency to impulsively overeat. Morbid obesity is worsened by decreased energy expenditure and reduced physical activity (Whitman et al., 2002).

PWS first appeared in the medical literature when endocrinologists Prader, Labhart, and Willi reported an unusual pattern of abnormalities, including diminished fetal activity, profound poor muscle tone, feeding problems in infancy, underdeveloped sex organs, short stature and retarded bone age, small hands and feet, delayed developmental milestones, characteristic faces, cognitive impairment, onset of gross obesity in early childhood due to insatiable hunger, and a tendency to develop diabetes mellitus (DM) in adolescence and adulthood when weight was not controlled. Butler and Thompson (2000), Cassidy and Schwartz (1998), and Cassidy and Schwartz (2009) have offered an expanded understanding of the syndrome.

Obesity and the concomitant development of DM have been of most concern for individuals with PWS. Dudley et al. (2008) conducted a cross-cultural comparison of body mass index and PWS. They discovered that obesity was similar for children with PWS in the USA, France, and Germany, but that French adults with PWS had significantly higher rates of obesity than adults in the USA or Germany.

Persons with PWS are at increased risk of experiencing emotional, behavioral, and cognitive problems (Whitman et al., 2002). Researchers have identified similar difficulties related to short stature among non-PWS individuals with growth hormone (GH) deficiency and idiopathic short stature. Moreover, normal adults with GH deficiency have reported a lower quality of life (QOL) and psychosocial problems.

During adolescence and adulthood, individuals with PWS develop psychiatric conditions, including acute cycloid psychosis, and obsessive–compulsive, bipolar, and pervasive developmental disorders (Descheemaeker et al., 2002). Persons with PWS may exhibit literal mindedness, inflexibility, and impaired social cognition (Whittington et al., 2004).

Growth hormone replacement therapy (GHRT) has produced positive changes in PWS children’s physical measures. In an investigation of adults with PWS, Hoybye et al. (2005) discovered that when GHRT was discontinued, the individuals exhibited impairments in both physical and social status and overall functioning.

GHRT has shown significant positive psychosocial and behavioral effects in non-PWS individuals (Whitman et al., 2002). In non-PWS populations, GHRT has enhanced alertness, physical activity, endurance, and extroversion. This therapy has
also reduced irritability and a person’s tendency to worry. These outcomes have improved relationships and reduced conflicts.

Based on a 2-year investigation, Whitman et al. (2002) discovered that GHRT significantly reduced symptoms of depression and did not result in a deterioration of behaviors during the treatment. The authors recommend using GHRT to treat PWS.

Bertella et al. (2007) assessed the QOL of those diagnosed with PWS who underwent growth hormone treatment. Results suggested that those with PWS had significant improvement with regard to both psychological and physical well-being.

**Angelman Syndrome (AS)**

When a deletion of chromosome 15 is found on the mother’s chromosome 15, the result is a syndrome different from PWS, known as Angelman syndrome (AS). AS appears to occur spontaneously for unknown reasons, as a result of deletion or disruption of a certain gene or genes on the long arm of chromosome 15 (15q11–q13). The syndrome was first identified by Angelman (1950), who described some children in his pediatric practice as having flat heads, jerky movements, protruding tongues, and having bouts of laughter (Angelman Syndrome Foundation, 2005; National Organization for Rare Disorders, 2003). The disease was initially referred to as the “happy puppet syndrome.”

AS is clinically characterized by severe mental retardation, lack of speech, EEG abnormalities, ataxia and stiff, atactic gait (Brouwer et al., 1990; Moncia et al., 1994; Robb et al., 1989). Other symptoms identified with AS include developmental delay, absence or near absence of speech, prolonged episodes of inappropriate laughter, characteristic facial features and episodes of seizures (Clayton-Smith and Laan, 2003; Garcia-Ramirez et al., 2008; Turchetti et al., 2006). Abnormalities of the head and face are typical, including microcephaly, deeply set eyes, macrostomia, maxillary hypoplasia, mandibular prognathism, and widely spaced teeth.

Infants with AS appear normal at birth, but often have feeding problems in the first few months of life. In addition, abnormal sleep patterns are typical. Furthermore, by early childhood, severe developmental delays are evident. Those with AS may also exhibit ataxia, resulting in a stiff manner of walking, with jerky arm movements and a characteristic positioning of the arms with flexion of the elbows and wrists.

Seizures often begin between 2 and 3 years of age. Although affected individuals may be unable to speak, many eventually learn to communicate through other means, particularly sign language. Others have adequate receptive language in order to respond to simple commands. The prevalence of autistic disorder is still debated (Pelc, Cheron, and Dan, 2008). Furthermore, Walz and Baranek (2006) confirmed a high degree and variety of sensory processing abnormalities in individuals with AS.

Although behavior problems have been documented in children with AS, researchers know little about their developmental course and outcome (Summers et al., 1995). Drawing on a review of case reports and parent responses to a
Patau Syndrome (PS)

Patau syndrome (PS), also known as trisomy 13, is a congenital disorder associated with the presence of an extra copy of chromosome 13 or a translocation of a portion of chromosome 13 (Patau et al., 1960). First identified by Dr. Klaus Patau, in 1960, the condition is also called Bartholin–Patau syndrome, named in part for Thomas Bartholin, a French physician, who described an infant with the syndrome in 1656.

The incidence of PS is approximately 1 per 12,000 live births (Baty et al., 1994b; Baty et al., 1994a; Delatycki and Garder, 1997).

Seventy-five to eighty percent of cases are caused by a trisomy, whether a full or partial trisomy. In the latter, physical symptoms tend to be less severe. In the remaining cases, there is an inherited translocation of chromosome 13. PS occurs in approximately 1 in 8,000–12,000 live births. Miscarriages frequently occur, while in other instances, stillbirths occur. As is typical with other trisomies, the risk of PS increases with the mother’s age, particularly if she is over 30. Gender, racial, or ethnic differences do not occur among individuals with PS.

According to Beers and Berkow (2004) and Best and Stallworth (2002), severity and symptoms vary from individual to individual, but are most severe when there is full trisomy 13. Holoprosencephaly and microcephaly usually occur. In addition, myelomeningocele may exist, where the spinal cord protrudes through a defect in the vertebrae of the spinal column. In addition, incomplete development of the optic and olfactory nerves usually accompany brain defects. The eyes may be unusually small or one eye may be missing. At times, the eyes are set close together or even fused into a single structure.

PS also results in individuals being born either partially or totally deaf. Many also have recurring ear infections. Furthermore, facial features appear flattened; the ears are malformed and low-set; and a cleft palate, cleft lip, or both are common. In addition, those with PS may have extra toes or fingers, permanently closed fingers, noticeably prominent heels, missing ribs, and loose folds of skin at the back of the neck.

Genital malformations are typical in those with PS, including ambiguous genitalia in males, an abnormally formed uterus in females, undescended testicles, and an abnormally developed scrotum.
In nearly all cases, the infant has respiratory problems and heart defects, including holes between the chambers of the heart, holes in the valves of the lungs and the heart, and malformed ducts that result in abnormal direction of blood flow and misplacement of the heart in the right side of the chest. Frequently, cysts develop in the kidneys and/or gastrointestinal system.

After the first month after birth, infants with PS are likely to experience feeding problems; reflux disease; constipation; high blood pressure; low muscle tone; ear, eye, and urinary tract infections; light sensitivity; irritability; scoliosis; and slow growth rate.

With partial PS of the distal segment, there is evidence of a distinctive facial appearance, with a short upturned nose, a longer than usual area between the upper lip and nose, and bushy eyebrows. Tumors on the forehead are also typical. With partial PS of the proximal segment, there may be a variety of facial features, including a receding jaw, large nose, and a short upper lip. Although partial PS results in severe mental retardation, these individuals typically live far beyond those with full PS.

Approximately 45% of PS babies die within 1 month after birth. As many as 70% die within the first 6 months. Survival into adulthood is very rare. However, Nanjiani, Hossain, and Mahgoub (2007) described a 51-year-old woman with PS.

Duarte et al. (2004) reported on a PS patient who had a long survival. They examined a 28-month-old girl who at birth was cyanotic, icteric, spastic, and who cried weakly. She had polydactyly in the left hand, congenital club foot and convex soles, and ocular hypertelorism. The girl also had a low nasal bridge, many hemangiomas throughout her body, cardiomegaly, and perimembranous interventricular communication. However, the patient did not have a cleft lip or palate. She had neuropsychomotor development retardation at birth, but had improved after physiotherapy and recreational treatments.

Edwards Syndrome (ES)

Edwards syndrome (ES) is another chromosomal disorder, specifically the result of an extra copy of chromosome 18. Also known as trisomy 18, ES affects approximately 1 in 3,000 live births and is the second most common trisomy. Data from the Support Organization for Trisomy 18, 13 and Related Disorders (SOFT) indicates that approximately 75–80% of newborns impacted by ES are female. As with other trisomies, as a woman becomes older, she has a greater risk of having a child with ES. Very rarely, a translocation may occur, resulting in a partial trisomy. As a result, the individual may have fewer and less pronounced symptoms.

Physical symptoms include clenched hands with index fingers overlapping other fingers, crossed legs, a hole, split, or cleft in the iris, low set and malformed ears, small jaw, small head, separation between the two sides of the rectus abdominis muscle, unusual shaped chest, underdeveloped fingernails, undescended testicles, and hernia. Abnormalities in the lungs, diaphragm, heart, and blood vessel formations are also common. In addition, the kidneys may be malformed. Furthermore,
the baby may have clubbed feet and either webbed or fused toes. Low birth weight is typical. Severe mental deficiency is expected as well.

In the most comprehensive study of trisomy 18, Naguib et al. (1999) evaluated 118 children diagnosed with ES. They reported that the majority of children died before the second week of life and that congenital heart and gastrointestinal abnormalities were the most prevalent medical issues. The investigators also reported that maternal age was associated with ES.

Double trisomy is rare. Tennakoon et al. (2008) reported on a male neonate with double trisomy (48XYY, +18), who was born to a 28-year-old gravida three, parity one mother at 35 weeks of gestation. The baby exhibited clinical characteristics of ES. The male neonate did not have a family history of DM and had not been exposed to chemicals. Discrepancy between genders may explain why the Y-chromosome is rarely involved in ES.

### Cri du Chat Syndrome (CdCS)

Cri du chat syndrome (CdCS), also known as 5-p syndrome and cat cry syndrome, is a relatively rare genetic disorder caused by the deletion of the p arm of chromosome 5. The cause is unknown. Most cases are not inherited. The deletion usually occurs as a random event. Individuals with CdCS typically have no history of the condition in their family. About 10% inherit the chromosome with a deleted segment from an unaffected parent who has a balanced translocation.

Mainardi (2006) reported that the incidence ranges from 1 in 15,000 to 1 in 50,000 live births. Distinctive features of CdCS include small head, broad nasal bridge, weak muscle tone, widely spaced eyes, and low birth weight. A high-pitched, cat-like cry is the most prominent clinical feature in the newborn and is typically diagnostic for the disorder. The high-pitched, cat-like cry has been localized to chromosome 5p15.3.

Symptoms vary from one individual to the next (Fang et al., 2008). The variability is usually related to different sizes and locations of deletions in chromosome 5p. Nonetheless, common clinical symptoms include a high-pitched “cat-like” cry, delayed development, difficulty with language, and mental retardation. Approximately 50% of the children with this syndrome learn sufficient verbal skills to communicate (National Human Genome Research Institute, 2008). Some individuals with CdCS learn to use short sentences, while others are restricted to a few basic words or gestures or sign language.

Using the Aberrant Behavior Checklist in a study of 146 persons with CdCS, Dykens and Clarke (1997) showed that hyperactivity was the most important and prevalent problem. In addition, individuals with the disorder exhibited aggressive behaviors, tantrums, stereotypic behaviors, and self-destructive behaviors. In persons with lower levels of adaptive and cognitive functioning and those who have been in medication trials, these difficulties were more evident. Symptoms of autism and social withdrawal were more common in those with translocations instead of deletions.
Clarke and Boer (1998) studied problem behaviors associated with CdCS, Prader–Willi, and Smith–Magenis. Using the Aberrant Behavior Checklist, the authors found that CdCS and the other two chromosome deletion disorders were related to more ratings of problem behaviors among individuals with these three chromosome deletion disorders than those in comparison groups.

Individuals with CdCS may engage in repetitive behavior. Moss et al. (2009) compared the prevalence and nature of repetitive behavior in genetic syndromes and found persons with this disorder exhibited unique patterns of repetitive behavior.

Researchers have investigated the behavioral aspects of CdCS during early childhood. Sarimski (2003) evaluated the early play behavior in children, aged 2–7, with the disorder and showed that these children were easily distracted and had a low rate of object-directed activities compared to two control groups. The author concluded that a low level of object-directed behaviors in young children may be early signs of hyperactivity, being easily distracted, and stereotypic behaviors, which are typical of the behavioral phenotype of older persons with CdCS.

Based on an investigation of a three-generation family with 5p terminal deletion (5p15.2-pter), Fang et al. (2008) discovered that mental symptoms varied within the family although the family members shared deletions of the same size. Two female family members with the condition showed moderate mental retardation and psychotic symptoms, such as persecution delusions, auditory hallucinations, self-talking, and self-laughing. In contrast, the other three male family members with the disorder did not exhibit any psychotic behaviors but did show evidence of mild to moderate mental retardation. The investigators suggest that other factors besides the size and location of 5p deletions may influence the development of mental symptoms in persons with this disorder.

Researchers have looked at factors that cause variability in mental retardation and related symptoms in persons with CdCS. Semaphorin F (SEMAF) and delta-catenin (CTNND2) are two genes that have been mapped to the “critical regions” (Cerrutti, 2006). These two genes may be involved in cerebral development and their deletion may cause mental retardation in individuals with CdCS. Phenotypic alterations in CdCS may result from the deletion of the telomerase reverse transcriptase (hTERT) gene, which is localized to 5p15.33.

Based on a report of a family of four who have a deletion slightly distal (6p15.3) to the critical region, Cornish et al. (1999) discovered that this family had only minimal cognitive impairment, indicating that persons who have deletions that only delete the distal critical region have milder intellectual disability and a much better prognosis than persons with the typical CdCS phenotype who can have profound learning impairment.

Most people with CdCS have normal life expectancies. Furthermore, individuals with this disorder are usually happy and friendly and appear to enjoy social interaction. However, those with severe behavioral problems can cause significant stress for their families.

Those with CdCS may also experience delays in walking, feeding problems, and scoliosis. A small percentage is born with serious organ defects and other life-threatening medical problems. Chang et al. (2007) reported that the most common
heart condition was atrial septal defect, followed by ventricular septal defect. They also reported that 21% had hearing impairments and that 34% suffered from airway problems.

Wolf–Hirschhorn Syndrome (WHS)

Wolf–Hirschhorn syndrome (WHS) is the result of a genetic error on chromosome 4. However, recent research (Bergemann, Cole, and Hirschhorn 2005; Zollino et al., 2008) suggests that the deletion alone is insufficient for the full development of the disorder and that the deletion of linked genes contributes to both the severity of core characteristics and the presence of additional syndrome problems. In 87% of WHS cases, the individuals do not have a family history of the disorder. The condition is caused by partial loss of material from the distal portion of the short arm of chromosome 4 (4p16.3) and is considered a contiguous gene syndrome. First described by Hirschorn and Cooper (1961) and Hirschhorn (2008), WHS affects females more frequently than males. Battaglia, Fillippi, and Carey (2008) indicated that there is a female predilection of 2:1. The disorder is estimated to occur in 1 in 20,000 to 1 in 50,000 births.

WSS causes malformations in most parts of the body due to the genetic error. Symptoms include distinctive facial features, including prominent forehead, wide set eyes, and broad beaked nose; all these features have been collectively described as “Greek warrior helmet” features. According to Heljic et al. (2004) other common characteristics include growth retardation, mental retardation, and midline fusion defects (cleft lip or palate and cardiac septal defects). Less commonly reported problems are skeletal abnormalities, coloboma iris, dysplastic kidneys, and agenesis of corpus callosum.

Based on an analysis of 80 WHS patients, Zollino et al. (2008) defined three different types of the WHS phenotype that generally were associated with the degree of the 4p deletion. One category consisted of a small deletion that did not exceed 3.5 Mb. This category is frequently correlated with a mild phenotype in which the individual does not have substantial malformations. The second and most frequent type consists of large deletions, with an average of 5–18 MB. These deletions produce the recognizable WHS phenotype. A very large deletion of more than 22–25 MB produces the third category. This third type represents the most severe phenotype and is not defined as typical WHS.

Recently, Fisch et al. (2008) investigated the cognitive-behavioral features of children, aged 4–17 years, with WHS. They found diverse cognitive-behavioral profiles. The investigators discovered that cognitive deficits in these children ranged from mild to severe. In terms of adaptive behavior, females showed slightly higher scores than males. The children showed relative strengths in verbal and quantitative reasoning as well as in socialization. Seven of the 12 children exhibited attention-deficit hyperactivity disorder. One child had symptoms suggestive of mild autism.
Seizures have also been reported as found in 50% of those with WHS (Chen, 2003). However, Battaglia et al. (2008) reported that 93% had a seizure disorder. In addition, low muscle tone; poor muscle development; short stature; malformations of hands and feet, chest, and spine; as well as malformations or underdevelopment of urinary and genital organs have been cited.

Children with WHS frequently have sleeping problems. Extinction of these sleeping disorders may be effective if they have been influenced and positively reinforced by their parents’ responses. One case study of a 6-year-old girl with WHS showed that extinction of sleeping problems was effective and was maintained during follow-up (Curfs et al., 1999).

Several factors predict prognosis of WHS children (Zollino et al., 2008). The degree of the deletion, the development of complex chromosome abnormalities, and the severity of seizures affect a person’s prognosis.

**Velocardiofacial Syndrome (VCFS)**

Velocardiofacial syndrome (VCFS), also known as 22q11.2 syndrome, Phelan–McDermid syndrome, DiGeorge sequence, conotruncal anomaly face syndrome, CATCH 22, Sedlackova syndrome, autosomal dominant Optiz G/BBB syndrome, or Cayler Cardiacal syndrome, is a chromosome microdeletion syndrome caused by microdeletion on chromosome 22. VCFS can result from simple deletion, translocation, ring chromosome, and less common structural changes affecting the long arm of chromosome 22, specifically the region containing the SHANK3 gene. VCFS is one of the most common genetic disorders and manifests in a variety of symptoms in multiple systems (Shprintzen, 2008; Hay, 2007). The disorder occurs with equal frequency in males and females and is underdiagnosed due to lack of clinical recognition and/or insufficient laboratory testing (Phelan, 2008). The condition affects about 1 in 2,000 to 1 in 4,000 newborns, although a number of researchers have argued that the incidence is higher (Phelan, 2008; Shprintzen, 2008).

The name, velocardiofacial syndrome, comes from the Latin words, velum, meaning palate, cardia, meaning heart, and facies, having to do with the heart. However, not all of these identifying features are necessarily evident in those with VCFS. According to Shprintzen (2008), VCFS has an expansive phenotype with more than 180 clinical features that involve essentially every organ and system. The author added that the syndrome has drawn much attention because a number of psychiatric illnesses, including attention deficit disorder, schizophrenia, and bipolar disorder, have phenotypic features similar to VCFS.

Symptoms of VCFS include cleft palate usually of the soft palate, heart problems, eye problems, feeding problems, middle ear infections, immune system problems, weak muscles, low calcium, scoliosis, tapered fingers, bony abnormalities in the neck or upper back, and distinctive facial features (elongated faces, almond-shaped eyes, long eyelashes, full cheeks, wide or bulbous nose, and unusual ears).

A report of a child with VCFS and deletion of 4q34.2 to 4qter revealed that a distal 4q deletion can produce a phenotype similar to VCFS when a 22q deletion
is not identified (Tsai et al., 1999). The authors recommend searching for other karyotype abnormalities when a VCFS-like phenotype is evident and a 22q deletion has not been found.

In an investigation of VCFS phenotype and deletion of 22q11.2 in Hungarian children, Morava et al. (2000) suggest that many children with VCFS may have different etiologies other than deletion of 22q11.2 even though the VCFS phenotype seems to be prevalent among Hungarian children.

Hercher and Bruenner (2008) evaluated patients with the disorder and found that there is a 25–30% risk of developing schizophrenia for those diagnosed with VCFS. They also postulated that there is an increased risk for other psychiatric illnesses, including bipolar disorder and schizoaffective disorder as well.

A longitudinal evaluation of adolescents with VCFS by Gothelf et al. (2007) revealed that sub-acute psychotic symptoms interacted with both the catechol O-methyltransferase (COMT) genotype and with baseline symptoms of anxiety or depression to predict 61% of the variability in psychosis severity during the follow-up period. The investigators conclude that treating VCFS children with sub-acute signs of psychosis and internalizing conditions, particularly anxiety, can reduce their risk of acquiring psychotic disorders in adolescence.

Gothelf et al. (1997) examined VCFS manifestations and microdeletions in schizophrenic patients and found documented hemizygosity of 22q11 in 3 out of 15 patients. They recommend that psychiatrists become more aware of the signs of VCFS in psychiatric patients so that further molecular studies can be conducted. They recommend screening suspected patients with a single marker such as D22S941 and examine further only those patients who have a single electrophoretic band.

Based on a sample of 326 patients in a Japanese psychiatric hospital, Sugama et al. (1999) identified 12 patients with minor facial dysmorphia. Chromosomal analysis with fluorescent in situ hybridization (FISH) was conducted in six patients who most likely had VCFS based on additional assessment. One of these patients, a 41-year-old woman, had chromosome 22q11.2 deletion. She was schizophrenic but had no substantial dysmorphia, such as cleft palate and cardiovascular anomalies. The authors suggest that psychiatric symptoms in VCFS can develop without major developmental abnormalities. Schizophrenic patients may have subtle aspects of VCFS, but go unrecognized in routine medical examinations.

Using a sample of patients with VCFS, Papolos et al. (1996) showed a strong relationship between VCFS and early-onset bipolar disorder. Their findings indicate that the microdeletion on chromosome 22q11 may be a risk factor for early-onset bipolar disorder.

About 65% of these children have a nonverbal learning disability, resulting in learning problems. When tested cognitively, they are found to have verbal IQ scores 10 points or greater than their performance IQ scores (National Human Genome Research Institute). Children with VCFS tend to have relative strength in reading, spelling, and rote memorization and relative weakness in math and abstract reasoning (De Smedt et al., 2003). Developmental delays are also very common.
Drawing on a sample of children and adolescents with VCFS, Oskarsdottir et al. (2005) demonstrated that children with this syndrome have various neurological, motor, and cognitive difficulties. Although the number and severity of their impairments vary, the combination of disabilities in these children leads to a low level of participation in activities.

Using a sample of 25 children, aged 6–12 years, with VCFS and 25 matched controls, De Smedt et al. (2009) showed that children with VCFS scored more poorly on number comparison but not on number reading. Children with VCFS performed worse on large addition and subtraction problems compared to controls. Their ability to perform backup strategies in addition and subtraction was worse than controls. However, children with VCFS preserved retrieval of arithmetic facts.

Individuals with deletion 22q13.3 syndrome may also display delay in language development (Benitez, 2009) and autistic-like behavior (Phelan, 2008). They often have problems with communication and social interaction.

In a longitudinal investigation of four children with VCFS, Scherer et al. (1999) showed that young children with the disorder have receptive-expressive language disability from the beginning of language acquisition. In addition, the children exhibited severely delayed speech and expressive language development beyond that predicted by their other developmental or receptive language performance. They had severely limited speech sound inventories and early vocabulary development, compared to children with cleft lip and palate and children with isolated cleft palate.

Based on an analysis of VCFS children of borderline or normal intelligence, De Smedt et al. (2003) discovered that the early academic achievement of VCFS children compared on average with their age-related peers. However, at an early age, individual children with VCFS showed wide variability in the domain of counting skills and mathematics and some already exhibited distinct learning impairments.

In a report comparing children with VCFS to those with cleft palate or velopharyngeal dysfunction (VPD), Baylis et al. (2008) demonstrated that the VCFS group had lower articulation functioning and nonverbal measures of intelligence than those with cleft palate or VPD. Speech perception did not vary significantly among the three groups. Articulation skills were associated with nonverbal intelligence and level of velopharyngeal dysfunction.

Researchers have demonstrated that younger children with VCFS exhibit more speech disability than older children with the syndrome or children with some of the phenotypic aspects of VCFS but who do not have the condition (D’Antonio et al., 2001). Younger children with VCFS exhibit smaller consonant inventories, more developmental mistakes, worse articulation disorder, and more glottal stop use. The investigators suggest that children with VCFS have speech production which is not only different from normal but also may be specific to the disorder itself.

Velopharyngeal insufficiency (VPI) occurs in about 70% of patients with VCFS because of cleft palate (Ysunza et al., 2009). VPI is much more prevalent because of abnormalities related to VCFS, including platybasia, hypotrophy of adenoid, enlarged tonsils, hypotonia, and abnormal pharyngeal muscles.
Based on an evaluation of 29 patients who had velopharyngeal surgery for correcting VPI, Ysunza et al. (2009) showed that tailor-made pharyngeal flaps were the strategy for treating VPI in VCFS patients. After undergoing a pharyngeal flap operation 17 (85%) patients developed normal nasal resonance or mild hypernasality. Four patients continued to have severe hypernasality after the procedure. No surgical complications occurred.

Widdershoven et al. (2008) examined the outcome of surgical correction of VPI in patients with VCFS and a control group consisting of patients without VCSF who had a palatal lengthening surgery. The control group improved their speech hypernasality more than the VCFS group. Acoustic nasometry outcomes did not vary between the VCFS and control groups. The researchers concluded that using palatal lengthening to treat VPI in children with VCFS is safe and efficacious. However, mechanical improvement is not associated with gains in speech for children with VCFS.

Musculoskeletal problems, such as scoliosis, can occur among children with VCFS. Morava et al. (2002) evaluated 20 patients for scoliosis and connective tissue problems who were consecutively diagnosed with VCFS and 22q11.2 deletion. They discovered that three of these children had substantial scoliosis and connective tissue problems. Two of these patients were thought to have possible Marfan syndrome and were referred to a genetics evaluation. The investigators recommend that scoliosis should be viewed as a prevalent condition in VCFS patients. In addition, they suggest that 22q11.2 deletion be a possible diagnosis in patients with unexplained scoliosis and developmental delay.

Turner Syndrome (TS)

Turner syndrome (TS) is a chromosomal disorder related to the x chromosome. In TS, the syndrome happens when one of the two x chromosomes normally found in women is missing or incomplete. TS alters development in females only and occurs in 1 in 2,500 female babies. However, the syndrome is much more common among miscarriages and stillbirths. Researchers have yet to determine which genes on the x chromosome are responsible for most signs and symptoms of TS. However, researchers have identified one gene, SHOX, which is important for bone growth and development. Missing one copy of this gene likely causes short stature and skeletal abnormalities. The condition is usually not inherited in families.

Females with this disorder tend to be shorter than average and are typically unable to conceive a child due to the absence of ovarian function even though the vagina and womb are totally normal. Other common symptoms include a webbed neck with folds of skin from the tops of shoulders to the sides of the neck, puffiness or swelling of hands and feet, skeletal abnormalities, and low hairline in the back and low-set ears. In addition, there is increased risk of heart defects and kidney problems. Furthermore, there is increased risk for high blood pressure, DM, osteoporosis, thyroid problems, and cataracts.
Other symptoms include an especially wide neck, arms that turn slightly out at the elbow, scoliosis, minor eye problems, a broad chest with widely spaced nipples, and a heart murmur, sometimes associated with narrowing of the aorta.

In early childhood, those with TS may have frequent middle ear infections, which can lead to hearing loss in some cases. During the age of puberty, girls do not begin to menstruate or develop breasts without hormone treatment at this time.

Children with TS are typically of normal intelligence and have good verbal skills as well as reading ability. However, some girls have difficulty with mathematics, memory functioning, and fine motor skills.

As with other abnormalities of sex chromosomes, TS has been associated with a greater incidence of neuropsychiatric disorders. Wustmann and Preuss (2009) reviewed existing reports on TS and psychosis, while Roser and Kawohl (2008) reported that TS occurs approximately threefold more frequently in female schizophrenics than compared to the general population. Finally, Marco and Skuse (2006) cited a link between the x chromosome and autism and evaluated the incidence of autism with TS and Klinefelter syndrome.

Growth hormone injections may be beneficial for some girls with TS, resulting in an increase in adult height by a few inches. Estrogen replacement therapy is often begun at the time of normal puberty, around 12 years of age, to facilitate breast development. Later, estrogen and progesterone are utilized to begin menstruation, in order to keep the womb healthy. Rubin (2008) cited the need for exogenous estrogen therapy to be initiated in coordination with the final phase of growth hormone therapy. Estrogen may also be helpful in preventing osteoporosis. Assisted reproduction techniques may assist some women with TS to become pregnant.

Klinefelter Syndrome (KS)

Klinefelter syndrome (KS), also known as an xxy male, is a condition that occurs in men as a result of an extra x chromosome. Occasionally some of the cells only have an extra chromosome, resulting in an xy/xxy mosaic. These latter individuals may have enough normally functioning cells to allow them to father children, although infertility is the most typical symptom in KS. The syndrome is found in 1 of 500–1,000 newborn males (National Institute of Child Health and Human Development, 2006). Woman who have pregnancies after age 35 have a slightly increased chance of having a child with KS.

Although the syndrome’s cause is common, the symptoms and characteristics that may result from having the extra chromosome is not common. In fact, many individuals never know that they have an additional chromosome.

Men with KS may have small, firm testes, a small penis, enlarged breasts, sparse facial, body and armpit hair, tall stature, long legs, and a short trunk. In addition, they are more likely than other boys to be overweight and tend to be taller than their fathers and male siblings.

KS has also been associated with an increased risk of breast cancer, extra-gonadal germ cell tumor, lung disease, osteoporosis, and varicose veins. Furthermore, those
with KS also have an increased risk for autoimmune disorders, including Sjogren’s syndrome and rheumatoid arthritis (Mazzocco & Ross, 2007).

Recent research has focused on hormonal and speratogenic testicular failure in those with KS. Although infertility has been cited as a primary problem, Paduch et al. (2008) found that over 50% of men with KS had sperm, and therefore were not sterile. However, recent evidence suggests that children with KS are born with spermatogonia and lose large numbers of germ cells during puberty. There has also been some evidence that those with KS have diminished sexual drive (Hunter, 1969), but hormone treatment has been effective in reducing this issue.

Although not mentally retarded, children with KS are often diagnosed with learning disabilities. Most have some degree of language impairment and exhibit problems with learning to read and write (Graham et al., 1988). In addition, they frequently have delayed speech development. The majority have some problem with language throughout their lives. Ross et al. (2008) assessed the neuropsychological functioning of children with KS and found that specific language, academic, attentional, and motor abilities were impaired. In the language domain, there was a relative deficit in higher linguistic competence, although vocabulary and meaningful language understanding abilities were generally intact. Deficits in the ability to sustain attention without impulsivity were also evident. Furthermore, children with KS demonstrated an array of motor problems, particularly in strength and running speed.

*xxy boys are usually well behaved in the classroom. They tend to be shy, quiet and want to please. When confronted with frustration academically, they tend to withdraw and daydream. Teachers sometimes fail to recognize the language problem and may dismiss them as lazy. As a result, they may fall further behind in school, and may eventually be held back. The risk of school failure and concomitant decreased self-esteem is prevalent. As a result, early identification is essential, with appropriate educational intervention.

The development of psychiatric problems has been widely discussed in the literature. For example, Polani (1969) reported that three times as many male schizophrenic patients had KS, as compared to the general population. Stuart, King, and Pai (2007) reported that autism spectrum disorders are heterogeneous with KS. However, Kessler and Moos (1973) indicated that there were no typical behavioral characteristics associated with a specific chromosomal disorder, including KS.

Developmental Disabilities (DD)

Frequently people with chromosomal abnormalities and other conditions, such as cerebral palsy and mental retardation, have developmental disabilities (DD). DD is synonymous with the terms learning disability (LD), intellectual disability (ID), and cognitive disability in certain countries.
DDs are usually classified as severe, profound, moderate, or mild, based on the individual’s assessed need for supports, which may be permanent. Many social, environmental, and physical factors cause DDs, although a definite cause for a person may never be determined. Abnormalities of chromosomes and genes, brain injury, extreme prematurity, child abuse, poor diet, and growth problems are common factors causing DDs.

Between 1 and 2% of the population in most western countries acquire DDs, although statistics are flawed in this area. About 1.4% of the worldwide proportion of people is thought to have DDs (Secretary of State, UK, for Health, March 2001). DDs are twice as common in males as in females. In areas of poverty and deprivation, the prevalence of mild DDs is thought to be higher, and among people of certain ethnicities, it is also higher.

Many people with DDs suffer from various physical health problems. Individuals with DDs have inheritable disorders such as DS. However, other major contributing factors are lack of access to health services and a lack of awareness by health-care professionals. Persons who have severe communication problems have a difficult time expressing their health needs and might not recognize their own health problems without adequate support and education. They may also suffer from sensory problems, vision and hearing problems, obesity, and poor dental health. Individuals with DDs have a life expectancy that is about 20 years below average, although with innovations in adaptive and medical technologies, they are living longer and healthier lives. Freeman–Sheldon syndrome and other specific diagnoses do not affect life expectancy.

Persons with DDs are more likely to have mental health problems and psychiatric disorders than those in the general population. Traumatic events, such as abuse, bullying, and harassment, social restrictions due to low socioeconomic status (SES), biological factors, such as brain injury and alcohol abuse, and developmental factors, such as a lack of understanding of social norms, are some of the factors that produce a high incidence rate of dual diagnoses in these individuals. The difficulties that health professionals have in diagnosing mental health problems and inappropriate treatment can worsen the mental health problems of persons with DDs.

People with DDs frequently suffer abuse, including physical abuse, neglect, sexual abuse, emotional or psychological abuse, financial abuse, legal or civil abuse, systemic abuse, and passive neglect. Their low SES, low self-advocacy skills, communication deficits, and lack of understanding of social norms increase their risk of abuse.

Peer abuse is a significant, if misunderstood, problem. In addition, persons with DDs have rates of crime that are also disproportionately high, and the criminal justice systems in many countries are not equipped to meet the needs of people with DDs as both offenders and victims of crime.

Some persons with DDs engage in challenging behavior, including self-injurious behavior, aggressive behavior, inappropriate sexual behavior, behavior directed at property, and stereotyped behaviors, such as repetitive rocking. A number of factors may cause these challenging behaviors in persons with DDs, including biological,
social, e.g., boredom, the insensitivity of staff and services to the individual’s wishes and needs, environmental, such as noise and lighting, and psychological factors, such as feeling excluded and lonely. Challenging behavior is often learned and rewarded. However, persons with DDs are often able to learn new, positive behaviors to achieve the same aims. The challenging behavior of individuals with DDs is frequently a reaction to the challenging environments that service professionals create for people with these conditions.

**Family Well-Being and Social Functioning**

Families who have children with chromosomal abnormalities and associated DDs can experience a significant amount of stress, decreased QOL, and impaired social functioning. Unfortunately, families have needs during the time of clinical assessments, but their needs are often neglected because of the child-centered focus of the clinical evaluations (Head and Abbeduto, 2007). Researchers have investigated the families of children with chromosomal abnormalities to determine which factors increase the risk of family stress and impairment. A family system approach is useful for analyzing how the well-being of family members can influence children with DDs (Head and Abbeduto, 2007). A family systems model can be employed to evaluate how child, parent, and family factors may affect the adjustment of children with chromosomal abnormalities and their siblings and parents. In addition, the belief systems of families with chromosomal abnormalities can influence the well-being and social functioning of the affected child and other family members.

Based on an investigation of siblings and parents of children with a disability, Giallo and Gavidia-Payne (2006) demonstrated that the level of risk for families and their degree of resilience correlated better with sibling adjustment than the sibling’s own stress and coping resources. The authors conclude that family and parental factors may affect sibling adjustment more than sibling resources and coping experiences, and interventions for siblings should incorporate these findings.

Sibling adjustment is often a life-long process. A sample of 54 siblings from two linked longitudinal investigations was used to assess sibling relationship for adults who have a brother or sister with ASD or DS (Ormond and Seltzer, 2007). The results indicated that siblings of DS adults had more contact with their brother or sister compared to siblings of ASD adults. Moreover, compared to the siblings of ASD adults, the siblings of DS adults indicated that they had higher levels of positive emotion in their relationship, felt more optimistic about the future for their brother or sister, and were less likely to note that their interactions with their parents had been altered. Siblings of DS adults had better sibling relations if the sibling had lower educational attainment, did not have children, lived in closer proximity to the brother or sister with DS, when the sibling relied on more problem-focused coping strategies, was less pessimistic about the future of the brother or sister, and when her/his life had been substantially affected by living with a brother or sister with DS.
Another investigation by Wang et al. (2007) demonstrated that DS children had better social adjustment than a mental age-matched group. However, the DS children had worse social adjustment than an age-matched group. They concluded that various factors, such as cognitive development, family factors, and newborn history, predict social adjustment of children with this disorder.

In a study of families of children with CdCS, Hodapp (2008) showed that the child’s degree of maladaptive behavior was the best indicator of family stress. The investigators also found that parents and the affected child’s siblings disagreed on the degree of interpersonal concerns among siblings. The siblings reported fewer interpersonal concerns compared to their parents who indicated that the siblings felt like they were being ignored and misunderstood.

In a longitudinal investigation of children with DS, Hauser-Cram et al. (2001) showed that the children’s type of disability was the best predictor of changes in maternal, but not paternal child-associated stress. The disability type of the children was also correlated with parent-associated stress. In addition to disability type, the children’s behavior problems and mastery motivation and other indicators of self-regulation and the mother–child interaction predicted parent well-being.

Families who have a child with a chromosomal abnormality often experience major life changes because of the consequences of coping with the child’s DDs. Using the results of focus groups, King et al. (2006) discovered through alterations in their views of the world, values, and priorities, parents of children with DS or autism can acquire a sense of coherence and control. Parents achieve coherence and control by changing the ways in which they view their child, their own role as parents, and the family’s role. Although families suffer the loss of goals and aspirations, they nevertheless cope with their child’s disability by altering their perceptions about life and disability and come to see the positive role of their children in the family.

The Health-Care Experiences of Families

Researchers have analyzed the ways in which clinicians can improve the health-care experiences of families who are given the diagnosis of chromosomal abnormality and related DDs. Drawing on an outpatient, tertiary-based model for parents of children with a newly diagnosed visual impairment and/or ophthalmic condition, Rahi et al. (2004) suggest that the most important needs are for information, particularly about education and social services, and emotional support from professionals, social networks (both formal and informal), and support groups. Multidisciplinary teams can coordinate health, educational, and social services for newly diagnosed children and their families.

Various factors can increase the satisfaction of families who receive a prenatal diagnosis of a chromosomal disorder. In an investigation of parent-reported experiences after receiving the prenatal diagnosis of ES, Walker et al. (2008) identified several aspects of health care that affect satisfaction with health care. Provider empathy, continuity of care, communication, valuing the fetus, and participation in
clinical decisions can affect parents’ satisfaction. The authors conclude that training, education, or team-based strategies can potentially modify these aspects of health care and improve parents’ satisfaction with their health-care experiences.

Maternal serum screening (MSS) can be effective in prenatal detection of fetal chromosome abnormalities (Hu, 2007). However, not all pregnant patients are offered MSS. A cross-sectional mailed survey of family physicians in Newfoundland and Labrador discovered more than 50% of the family physicians provided MSS to all of their pregnant patients (Cavanagh and Mathews, 2006). Another third of the family physicians surveyed offered it to some of their pregnant patients. In their study, the family physicians’ practice characteristics were not associated with their attitudes toward MSS or knowledge about it. Other factors besides physician practice characteristics should be considered in developing interventions to expand the use of MSS.

Understanding women’s decision to get MSS can help health-care providers expand their use of MSS. Using qualitative interviews with pregnant women, Park and Mathews (2009) demonstrated that women are more likely to put greater emphasis on information obtained from informal sources (e.g., family, friends, and the Internet) than clinicians and researchers who are more likely to rely on information obtained through rigorous, peer-review research.

Improving families’ health-care experiences also can be enhanced through the use of genetic ultrasound. For example, investigations have found that second-trimester genetic sonography may be useful for identifying fetuses at risk for DS (DeVore, 2003). For high-risk women, genetic sonography is an alternative to universal amniocentesis. Genetic sonography reduces the loss rate of normal fetuses, which are subjected to amniocentesis because of risk factors related to advanced maternal age or abnormal maternal serum screening.

Drawing on Monte Carlo modeling, Benn and Egan (2008) reported that multi-step screening protocols should be used to maximize the advantages of both MSS and ultrasound and minimize the amount of testing provided.

School Programs for Children with Chromosomal Abnormalities

Educational training can be beneficial for children with chromosomal abnormalities. However, many families do not have access to or participate in these training programs. Even if their children do attend these special education programs, they may not receive the appropriate interventions. In addition, more research is needed to determine the effectiveness of special education programs in improving the functioning of children and adolescents with chromosomal abnormalities.

An investigation by Jaruratanasirikul et al. (2004) evaluated a school program for DS children in southern Thailand. Among these children, congenital heart disease and gastrointestinal anomalies were prevalent. The mortality rate of these children was 13.2%. The authors discovered that a majority of the DS children (65.6%) participated in an early intervention program. However, only 38.9% received a speech
intervention program. Family income predicted school attendance. Researchers are still following only 67.9% of the DS children aged over 5 years.

Researchers are examining the evolution of behavioral and psychiatric symptoms of children with chromosomal abnormalities from their primary school education to adulthood. Descheemaeker et al. (2002) analyzed the changes in behavioral and psychiatric symptoms among children, adolescents, and adults with PWS. They discovered that PWS individuals who were regarded as active and extrovert toddlers and who exhibited autistic symptoms in primary school later had psychotic episodes. These PWS individuals had moderate to severe mental retardation. In contrast, PWS persons who were considered passive and introverted as toddlers and exhibited less impaired behavior during their primary school education later had an unstable mood disorder. These PWS individuals’ intellectual functioning was in the normal to borderline range.

Children with chromosomal abnormalities may have difficulty achieving their full intellectual potential in school. A variety of factors may contribute to their academic underachievement. Whittington et al. (2004) analyzed the school performance of a group of children with PWS and a comparison group of children with LD. They discovered that both groups of children had lower levels of achievement than would have been expected based on their IQ scores. Some children with PWS, however, did function as expected in at least one academic domain. Among PWS children, academic underachievement in different domains was positively associated with the amount of time spent in special school.

The authors suggest that children with PWS may be placed in special education programs in large part because of their behavioral difficulties or physical impairments or based on expectations about their disorder (Whittington et al., 2004). PWS children’s impaired social behaviors may in fact cover up their true academic abilities. Interventions should take into account their behavioral problems and poor socialization skills, which may be masking their true intellectual ability.

Case Study

The following case study describes a girl with PWS. Janis is a 12-year-old Caucasian female, born to upper-middle-class parents. She has one older brother, 16 years old. She exhibited feeding problems in infancy and very poor muscle tone. The pediatrician showed no concern when the parents first expressed concern. When she was approximately 2.5 years, she began to eat excessively and exhibited pica, for example, eating dirt from plants in the home. The parents then went to a pediatrician affiliated with a children’s hospital and Janis was subsequently evaluated by a geneticist and diagnosed with PWS.

She was initially sent to an early childhood education program, but her behavior was so disruptive to the other children that Janis was referred to another program for children with severe emotional and behavioral problems. At home, her behavior was also a problem, including impulsive eating and other impulsive behavior. Janis would periodically ruin her older sibling’s clothes, electronics, and school work.
Eventually, a lock had to be placed on each room as well as the refrigerator, pantry, and cabinets.

Her parents had increasing conflict as Janis became older. Eventually, the child’s father threatened divorce unless Janis was institutionalized. When Janis began to smear feces in the house, the mother agreed to placement. Janis has been placed in a private facility since 10 years old and goes home on weekends for visits. She has adjusted well to the private placement.

Janis’s parents have also worked with a behavior therapist, in order to learn skills in helping ameliorate their daughter’s behavior when she is home. The focus has been on replicating the approach utilized at the group home. Low doses of risperdal have also been utilized to control impulsive behavior.

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