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An Exploration of the Cognitive, Physical and Psychosocial Development of Children with Apert Syndrome

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ABSTRACT

Apert syndrome is a rare condition, with a birth prevalence of approximately one in 65,000. This article provides an up-to-date review of the literature on Apert syndrome from a variety of perspectives, ranging from surgical management to personal accounts. The purpose of the review is to provide a holistic description of the syndrome which should be of assistance to those interested in understanding the impact of Apert syndrome on children and young people with the syndrome. Children with Apert syndrome are at risk for a range of impairments, disabilities and consequent complex needs which can have implications for their personal development, social inclusion and education. As a consequence, those involved in the care of children and young people with Apert syndrome have the challenging task of balancing the management of surgical interventions alongside the needs of the growing child.

KEYWORDS

Apert syndrome; craniosynostosis; FGFR2; hand anomaly; hearing impairment; social inclusion; syndactyly; visual impairment

Introduction

Apert syndrome is a rare syndrome which was first described by Wheaton in 1894, and investigated further by Apert in 1906 (Patton, Goodship, Hayward, & Lansdown, 1988). One of the largest studies, carried out between 1983 and 1993 in the USA, estimated a birth prevalence of Apert syndrome of approximately 1 in 65,000, in North America and Europe (Cohen et al., 1992; Tolarova, Harris, Ordway, & Vargervik, 1997). Lajeunie et al. (1999) suggested that this number may now have reduced due to improved diagnostic facilities and parents' decisions to terminate pregnancies. In Apert syndrome, the cranial (and facial) sutures begin to fuse early (craniosynostosis) during foetal development. This early fusion alters the normal pattern of skull growth and therefore the shape of the skull and face, which can raise pressure within the skull (intracranial pressure) and have consequences for development of the brain. The rate and order in which different skull sutures fuse determine how the skull shape is affected. Children with Apert syndrome are also born with their fingers and toes fused (syndactyly). Using ultrasound during pregnancy, it is possible to observe ventriculomegaly at 22 weeks, syndactyly of the fingers and toes at 25 weeks and an unusual skull shape at 28 weeks (Respondek-Liberska, Smigiel, Zielinski, & Sasiadek, 2010).

Apert syndrome is caused by a mutation of the fibroblast growth factor receptor 2 (FGFR2) gene (Wilkie et al., 1995). There are two adjacent mutations, called S252 W and P253R, which

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account for 98% of cases of Apert syndrome (Wilkie et al.). Wilkie and colleagues suggested that the two variants of Apert syndrome could be distinguished by the severity of the syndactyly of the fingers. In a study of 40 patients with Apert syndrome, they found that the S252 W mutation was more common and could be distinguished from the P253R mutation by the less complex syndactyly of the fingers, but often more severe craniosynostosis. Slaney et al. (1996), however, suggested that this classification was not so clear cut and could depend on what classification strategies were used for the hands. Lajeunie et al. (1999), in a study of 36 patients with Apert syndrome, found that 8 out of 12 patients with the P253R variant had severe syndactyly, compared to 6 out of 23 with the S252 W variant. More recently, Fearon and Podner (2013), in a review of 135 patients with Apert syndrome treated in a specialist unit, over a period of 20 years, also found that patients with the S252 W variant were more likely to have less severe syndactyly than patients with the P253R variant. In Fearon and Podner's review, only patients with the S252 W variant had a cleft palate.

Much of the literature on Apert syndrome focuses on the management of the syndrome from a clinical perspective. While there is much in the medical literature about the range of surgical interventions and optimal times for carrying out this surgery, much less is known about the cognitive and social development of children with Apert syndrome.

As in the general population, the cognitive development of children born with Apert syndrome varies. There are claims that children with Apert syndrome present with below average IQ (intelligence quotient) scores. For example, in a retrospective study of the records of 29 patients with Apert syndrome, carried out in 1988, IQ scores were >70 in 48%, 50–70 in 31%, 35–49 in 14% and <35 in 7% (Patton et al., 1988). In a more recent study by Lajeunie et al. (1999), out of 24 patients with Apert syndrome, 15 had an IQ score of less than 70, with most of these being in patients with the S252 W variant. In the general population, the majority of IQs are in the range 85–115.

Trying to locate a cause for these deficits reveals the complex nature of the profiles of children with Apert syndrome, which may include visual and hearing impairment as well as difficulties with manipulating objects. It is therefore unlikely that the conventional psychometric tests are able to provide reliable data for children with Apert syndrome. Consequently, the early research, much of which suggested that the majority of children with Apert syndrome have learning difficulties, has been questioned (Shipster, Hearst, Dockrell, Kilby, & Hayward, 2002). More recently, it has been suggested that there may be a relationship between the number of operations performed on a child and later developmental outcomes (Fearon & Podner, 2013). When analysing their results, Fearon and Podner were surprised to discover the high number of surgical procedures that were performed on the patients in their study. Their results showed that at a mean age of eight years, most patients had undergone 10 surgical procedures. In addition, their results showed a correlation between high numbers of surgical procedures and lower levels of neurocognitive development. However, the strongest correlation was between higher cognitive levels and medical treatment from only one centre. Fearon and Podner argue that through coordinating procedures more carefully and ensuring a minimum number of surgical operations in young children, developmental outcomes might be improved for children with Apert syndrome.

This literature review should serve to provide an overview of the key issues raised, in relation to understanding, managing and supporting children with Apert syndrome. In order to provide a review which represents the majority of experiences of Apert syndrome, the review has, as far as possible, focused on large studies rather than reports of individuals. The

review was undertaken using the author's institutional search facility which included Pubmed, Ovid and Scopus (Elsevier). This was then supplemented using Google Scholar for further readings. In addition, bibliographies were used to ensure the use of primary sources, as far as possible. Publications were limited to those available in English. Searches were initiated using the following text words and phrases: Apert, Apert syndrome, Apert cleft, Apert hands, Apert hearing, Apert speech, Apert vision, craniosynostosis brain, and midface advancement, white matter craniosynostosis.

The Cognitive, Physical and Psychosocial Development of Children with Apert Syndrome: What is typical?

An overview of some of the most significant factors which impact the development of children with Apert syndrome will be described. This is important because, although children with Apert syndrome show substantial variation in cognitive development and a wide range of IQ scores, it has not been possible to identify a cause for this variability (e.g. Lefebvre, Travis, Arndt, & Munro, 1986; Patton et al., 1988; Renier et al., 1996; Yacubian-Fernandes et al., 2004). Issues discussed in previous research, which are thought to have the potential to impact the cognitive development of children with Apert syndrome, have included:

- Age of children when first cranial surgery is performed
- White matter anomalies, such as ventriculomegaly, anomalies of the septum pellucidum or anomalies of the corpus callosum
- Visual impairments and hearing impairments, usually due to the premature fusion of the bones in the skull
- Poor fine motor skills due to fusion of the fingers
- Speech and language development which may be due to the children's unusual oral structures and frequent hearing impairments
- Psychosocial aspects and home environment.

Some of these issues will be discussed further in order to better understand the challenges faced by children and young people with Apert syndrome.

Cranial Surgery and Cognitive Development

There is much discussion about the possible link between cognitive development and the age at which children undergo their first cranial surgery. Renier et al. (1996) found that cognitive development was better in children who had undergone cranial surgery in their first year of life. Others, however, report that there is no correlation between IQ and the time of first cranial surgery (e.g. Patton et al., 1988). In a more recent study (De Jong, Rijken, Lequin, van Veelen, & Mathijssen, 2012), it was found that children with syndromic craniosynostosis (including Apert syndrome) usually have a normal or enlarged intracranial volume, even without surgery. This, they suggest, might be caused by the excess of cerebrospinal fluid which forces the skull to grow in order to compensate for the prematurely closed sutures. In this way, the brain is allowed to develop normally, even inside a skull which may be unusual in terms of its shape and structure. While Renier et al. identified some unusual structures in the brains of some of the children with Apert syndrome, these were specific cases, rather than generalised patterns of development. In most of the studies undertaken, sample sizes

were small and where larger samples were used, the data analysis is often based on retrospective data from hospital records (e.g. Patton et al.).

White Matter Anomalies

There have been conflicting findings in relation to the role of white matter anomalies in determining developmental outcomes for children with Apert syndrome. A number of studies (e.g. Cohen & Kreiborg, 1990; Quintero-Rivera et al., 2006) have concluded that children with Apert syndrome are at greater risk for hypoplasia of white matter and malformations of the corpus callosum, septum pellucidum and the limbic system. It has been suggested that these malformations may put some children with Apert syndrome at greater risk of having problems with memory, cognitive function and behaviour. It has also been suggested that children with anomalies of the septum pellucidum are more likely to have an IQ < 70 (Renier et al., 1996). This conclusion, however, must be approached with caution, as there is not yet sufficient evidence to conclude that these malformations cause problems with cognitive function (Cohen and Kreiborg).

In addition, these studies need to be contrasted with the work of Fearon and Podner (2013), who did not find any correlation between cognitive development and either anomalies of the septum pellucidum or anomalies of the corpus callosum. Moreover, it has been suggested that although there may be changes in white matter and other aspects of brain development in children with Apert syndrome (Raybaud & Di Rocco, 2007; Stark et al., 2015), it is hard to identify a causal link between these changes and neuropsychological outcomes (Florisson et al., 2011). This is an area that may still benefit from further study both within the literature on Apert syndrome and in the literature on the role of white matter more generally.

Visual Impairments in Children with Apert Syndrome

Children with Apert syndrome often experience a range of problems with their eyes and, consequently, with their vision. These 'result from disproportionate growth of the brain and eye. Premature closure of multiple cranial sutures results in the restriction of intracranial and orbital space expansion' (Khong et al., 2006, p. 349), so that the eye sockets are not able to grow to full size. The position of each of the eyes on the face may also be affected by the premature closing of sutures in the face.

Children with Apert syndrome always have hypertelorism. As a consequence, they are more likely have a squint and other related problems with their vision because their eyes are far apart (Kreiborg & Cohen, 2010). Children with Apert syndrome are often born with eye muscle anomalies, including missing or underactive muscles and there may also be blocking of the tear ducts (Jadico et al., 2006). The fact that their eyes may not close fully when they sleep may cause children with Apert syndrome to have scarring of the cornea (Khong et al., 2006).

A review of hospital records of 37 patients with Apert syndrome, managed by the Australian Craniofacial Unit from 1975 to 2004, identified 69% with some form of ametropia (Khong et al., 2006). These are difficulties which could include being long-sighted or short-sighted and having astigmatism. An important finding of this study was that even when children with Apert syndrome were given glasses to correct for their visual difficulties, many

of them did not have visual acuity within the normal range. The results for corrected visual acuity (i.e. with glasses) showed that 54% of the children had a corrected visual acuity of less than 6/12 in at least one eye and 17% had a corrected visual acuity of less than 6/12 in their best eye. A visual acuity of 6/12 is a way of defining vision by comparing the vision of the affected person with what is considered to be 'normal' vision. A corrected visual acuity is the visual acuity of the affected person when they are wearing glasses. If the visual acuity obtained with glasses is not within normal limits, there will be other problems with the eye which means that it is not possible to provide 'normal' vision. A corrected visual acuity of 6/12 means that, with glasses, the affected person sees at 6 metres what a person with normal vision sees at 12 metres. This could make things like copying from a board and playing football or tennis quite challenging. In addition to these difficulties, 13 of the patients had amblyopia (having eyes that do not work together), which in Apert syndrome is usually caused by strabismus (squint) which, when combined with other unusual structures, can cause a lack of stereoscopic vision and, consequently, an inability to judge distances effectively (Read, 2014).

A study of eight children with Apert syndrome attending Great Ormond Street Hospital, in London, UK, showed that all the children had mild or moderate visual field loss (this is usually described in terms of the angle the eye can see towards the nose, away from the nose and above and below the midline) in both eyes (Liasis et al., 2011). The reasons for this are not clear, but it has been suggested that it might be due to damage caused by raised intracranial pressure during sleep, itself caused by persistent obstructive sleep apnoea.

Children with Apert syndrome, then, are likely to be affected by some form of visual impairment. This could affect their ability to judge distances and speed (which could impact their ability to join in with games such as football and netball). A visual impairment could also impact a child's ability to scan and track information presented visually. A visual impairment might also mean that even if a child can read, they might need to have texts available in larger fonts with a high quality of production, rather than poor-quality, enlarged photocopies.

Hearing Impairments in Children with Apert Syndrome

Acquired hearing loss is common in children with Apert syndrome (Rajenderkumar, Bamiou, & Sirimanna, 2005a). This is attributed to poor functioning of the Eustachian tubes as a consequence of which children with Apert syndrome often suffer from glue ear (otitis media with effusion) or the consequences of glue ear (e.g. perforated ear drums) into adulthood. Of the 34 children over the age of 10 years who were patients at Great Ormond Street Hospital, in London, UK, between January 1970 and September 2003, 19 children developed conductive hearing loss which was either mild or moderate; only eight did not develop any persistent hearing loss. In a review of all 70 children who were patients in this period, 65 had at least one documented episode of glue ear and 3 of those that had no reported incidence were under four years of age. It was also noted that the acquired hearing loss experienced by these children tended to affect the lower frequencies (Rajenderkumar, Bamiou, & Sirimanna, 2005b) so that children were able to hear high pitch noises better than low pitch noises. As human speech is often within the range of lower pitch sounds, this type of hearing loss may affect children's ability to engage in conversation and interact appropriately.

Rajenderkumar et al. (2005a) recognise that there is a need to work out the best way of optimising hearing levels and managing problems associated with glue ear.

Zhou, Schwartz, and Gopen (2009) reviewed the records of 20 children who had all been patients at the Children's Hospital Boston, USA. Eighteen of the children had hearing loss, but analysis of their CT scans revealed air bone gaps and a number of structural anomalies of the inner ear (typically in the cochlea, semi-circular canals and the vestibule), in addition to the otitis media with effusion discussed above. The role of these anomalies is currently not well understood, but it is argued that they could have a role in causing the hearing loss in children with Apert syndrome.

As a consequence of the high incidence of persistent hearing loss, children with Apert syndrome are at risk of developmental delays in speech, language and communication (Deafness Research UK, 2009). This can result in social and behavioural problems and gaps in general knowledge, in addition to delays in learning to read and the development of reasoning skills.

Children with hearing impairments are likely to have delayed phonological development (Shipster et al., 2002). This might not only have an impact on their speech and language development, but could also have an impact on their phonological working memory and make the acquisition of reading and any other learning which requires the processing of aural information particularly challenging.

Children who experience hearing loss are at risk of missing key bits of information during activities in school which could make it very difficult when new learning is built on existing learning. If the child does not hear a key piece of information, then any subsequent work will not make sense. In addition, a hearing impairment might make the learning of new vocabulary difficult because the new words will be missed, or will not be heard correctly. For a young child, for example, this could cause a lot of confusion when learning number words, where it might be hard to distinguish between words such as 'thirteen' and 'thirty'. Finally, if a child has a hearing impairment, the very act of listening will be much more tiring than for a child without a hearing impairment. The consequence of this could be that a child with a hearing impairment simply 'switches off'.

Hand Anomalies and Outcomes for Children with Apert Syndrome

Children with Apert syndrome are always born with some fusion of the fingers. These can be categorised into three types:

- Type 1 is syndactyly of the three middle fingers (digits 2 to 4)
- Type 2 is syndactyly of all the fingers (digits 2 to 5)
- Type 3 is syndactyly of all five digits. (Cohen & Kreiborg, 1995)

In other words, the index, middle and 'ring' fingers are always fused in children with Apert syndrome, with boney fusion of the tips of the fingers. The 'little' finger is usually, but not always, fused. The last joint on the little finger usually bends. The thumb is usually short and wide and it bends outwards. This makes it very difficult for children to have a 'normal' pinch grip, even after surgery.

Surgery is usually performed in stages before a child reaches five years of age. Depending on the severity and complexity of the fusion, children usually have either three or four fingers and a thumb following surgery. In an investigation of an adult with Apert syndrome, prior

to surgical separation, the fingers were represented in the brain, as one single digit (Mogilner et al., 1993). Within one week after surgery, the hand area in the brain had increased and the fingers had more distinct cortical representation locations. This was followed up for six weeks after surgery and the changes remained stable. However, 'the resulting hand area was smaller than normal and the organization was nonsomatotopic' (Mogilner et al., p. 3597). This suggests that, in addition to the cosmetic benefits and greater dexterity, separation of digits results in a changed cortical representation of the hand. Nevertheless, the resulting representation of the fingers in the brain will not be the same as that in individuals with a 'normal' hand which may be partly explained by the fact that the new fingers will not have 'normal' pads (Mogilner et al.).

To make things more complicated, children with Apert syndrome often present with additional abnormalities with bones, cartilage, muscle and neurovascular development (Salazard, Galinier, Samson, Casanova, and Magalon (2007)). Surgical management is concerned with optimising the function as early as possible to ensure cortical integration. Children with Apert syndrome have interphalangeal joints that do not work properly, and they are, therefore unable to bend any fingers, except, sometimes, the tip of the fifth digit.

Salazard et al. (2007) reported on the results of surgery on 16 hands of children with Apert syndrome. Of these hands, seven had five digits and nine had four digits. Each child had, on average, six operations in total. On completion of the surgery, all the children had a functional pinch grip, which made it possible to hold a pen, but had difficulty making a palm grip so that things like using the brakes on a bicycle and doing up buttons were either difficult or impossible.

In addition to its functional usefulness, the hand plays an important role in relation to sensory input and communication (Andersson, Gillberg, Fernell, Johansson, & Nachemson, 2011). The hand is difficult to hide and may impact on the well-being and self-concept of those with hand anomalies. Andersson et al. found that boys with hand anomalies considered themselves to be significantly less popular than boys with no hand anomalies. It is possible, then, that children with Apert syndrome may be highly self-conscious about their hands. This might impact on their willingness to use their hands and fingers in school and in public places.

Speech, Language and Cognitive Development

Speech development in children with Apert syndrome may be affected by a cleft palate and/or midface advancement. A cleft palate in a child without Apert syndrome will cause delayed acquisition of speech sounds (Chapman, Hardin-Jones, & Halter, 2003). A child with Apert syndrome and a cleft palate will have to deal with the challenges of a cleft palate, in addition to the challenges that result from the other unusual oral structures that are due to the syndrome. In a study involving four patients with Apert syndrome, but no cleft palate, in London, UK, midface advancement was found to improve speech in all cases (Pereira, Sell, Ponniah, Evans, & Dunaway, 2008). These findings, however, were not supported by a retrospective study of resonance and speech following midface advancement involving seven children with Apert syndrome in Liverpool, UK (Bordbar, Blumenow, Duncan, & Richardson, 2012). Of the four children with a cleft palate, three experienced deterioration in their velopharyngeal function following midface advancement and two of those had later surgery to improve their speech (there was no information here on when, or even if, the children had had their

cleft palates surgically closed). Of the three children without a cleft palate, two experienced deterioration in their velopharyngeal function following midface advancement and both of these also had later surgery to improve their speech. It seems that the impact of a cleft palate and of midface advancement on speech is an area that would benefit from further exploration.

A study of 10 children with Apert syndrome, in London UK, between the ages of four and five years revealed that 8 out of the 10 children had language difficulties, six of these children performing better on tests for receptive language than for expressive language (Shipster et al., 2002). British Ability Scales (BAS) II were used to assess cognitive skills and the results were analysed using General Cognitive Ability (GCA) and Non Verbal Composite (NVC). It was found that there was no significant difference between GCA and NVC scores and all the children achieved a NVC score within the normal range. It is important to note the fact that this study used the British Ability Scales which are less demanding in terms of attention and fine motor skills than other assessments such as the Wechsler Intelligence Scale (Maliepaard, Mathijssen, Oosterlaan, & Okkerse, 2014). This might help explain the findings of Lefebvre et al. (1986) who reported that children with Apert syndrome had lower verbal intelligence scores than their overall performance scores (using the Wechsler Intelligence Scale for Children (WISC)). All the children in Shipster et al.'s study had hearing difficulties, with some of them having histories of mild to moderate conductive hearing loss. The children also all had articulation problems with their speech due to their unusual oral structures. As a consequence of these factors, it is not surprising that the children exhibited problems with speech and language development.

Difficulties with attention control were identified in 9 of the 10 children studied by Shipster et al. (2002). The authors point out that it is not unusual for children with speech and language difficulties to have problems with attention control. The authors also reported that all the children had phonological delays which, if not identified and addressed, could impact the children's development of literacy skills.

As already mentioned, early studies of the cognitive development of children with Apert syndrome reported a high incidence of lower IQ scores than in the general population (e.g. Lajeunie et al., 1999; Maliepaard et al., 2014; Patton et al., 1988). In a more recent study of 18 people with Apert syndrome (Yacubian-Fernandes et al., 2004) aged between one year and 26 years, with an average of eight years, assessments on developmental or cognitive functions were correlated with the age of the patient, age at which cranial surgery was performed, brain development and quality of family environment. In this study, 77.8% of the children had cognitive assessment scores within the normal range. The strongest correlation was between cognitive assessment scores and home environment (based on number of family members, level of education, type of accommodation, job and salary). Another study, by Renier et al. (1996), found that the children who had grown up with their families were three times more likely to have a normal IQ when compared with children who had been institutionalised at an early age. This suggests that the environment in which children with Apert syndrome grow up is a significant factor in their later development.

Psychosocial Development and Social Inclusion

De Jong, Maliepaard, Bannink, Raat, and Mathijssen (2012) reporting on a study of 131 children (20 with Apert syndrome) aged between 4 and 18 years found that the overall quality

of life for children with syndromic craniosynostosis was lower than for typically developing children.

Sarimski (2001) identified the fact that peer relationships for children with Apert syndrome are often unsuccessful and that children often experience rejection and hostility. This may cause children to become withdrawn and anxious in social situations in order to avoid behaviours such as name calling, staring and pitying. These children are, therefore, more likely to opt out of activities with peers in school and in social situations more generally.

In a study focusing more specifically on behavioural and emotional functioning of children with craniofacial syndromes (including six children with Apert syndrome), Maliepaard et al. (2014) found that children with syndromic craniosynostosis were more likely to have social problems and internalising problems. This could mean that children with Apert syndrome are more at risk for social isolation, poor self-esteem and depression. According to Maliepaard et al., children with syndromic craniosynostosis are also more at risk for attention problems, including ADHD (Attention Deficit and Hyperactivity Disorder). Additionally, it seems that the children who are most likely to have emotional and behavioural issues are those with lower scores for intellectual functioning.

Given the fact that Apert syndrome affects facial and physical appearance, this aspect is worth some discussion. Although there is only limited information available on this, a study by Shapiro, Waljee, Ranganathan, Buchman, and Warschausky (2015), exploring attitudes to appearance of parents of, and children with craniofacial anomalies, identified that parent and child ratings for appearance were not correlated, especially for girls. While there are more pressures on girls to conform to certain social norms, it seems that girls with craniofacial anomalies are more likely than boys to suffer from social isolation and anxiety as a result of their visible difference than their male peers.

In the study conducted by Shipster et al. (2002), parents of children with Apert syndrome reported concerns about their children's psychosocial inclusion at school and in the wider community. They also reported that their children were far less communicative with people outside the home, especially in group situations and at school. This could result in delayed development of social skills, as well as speech, language and communication skills. This could then be followed by lower expectations of children in school which could impact children's self-image, goals and expectations.

In the school environment, the issue of social inclusion is important. Children with a visible difference (disfigurement), and unusual speech sounds, are more likely to experience bullying at school than their peers (Turner, Thomas, Dowell, & Rumsey, 1997). It is not uncommon for people with a visible difference to have difficulties in social situations, have low self-esteem, be fearful of the negative reactions of others and have negative patterns of behaviour, such as avoiding social interactions (Rumsey & Harcourt, 2004).

Children with Apert syndrome are clearly likely to benefit from environments that are well-informed, supportive and accepting of them in order to support them to build their self-confidence and a positive self-image in relationships outside the family.

Personal Experiences

A review of this kind would not be complete without some consideration of the reflections and experiences of those experiencing Apert syndrome first-hand.

Living with Apert Syndrome

In 2012, Christine Clinton, a young woman with Apert syndrome, published a book entitled *'My Life Story with Apert Syndrome in My Own Words'*. In her reflections, Clinton discusses her childhood experiences which included: endless teasing and bullying at school; problems with friendships; low expectations from teachers; and a general lack of 'acceptance and tolerance'. Clinton highlights the fact that her positive experiences in education seemed to depend on individual teachers and their attitudes towards her—the positive experiences were the exception rather than the norm. This discussion takes place alongside descriptions of frequent hospital visits and surgical procedures. Throughout the book, Clinton emphasises the importance of family love and support.

Parents' Experiences of Raising Children with Apert Syndrome

In a study of mothers' experiences of bringing up a child with a craniofacial anomaly, one of the main concerns of mothers of children with Apert syndrome was to protect their child from the impact of negative attitudes and behaviours (Klein, Pope, Getahun, & Thompson, 2006). One mother talking about a trip to a ball game reported that 'Usually it's really fun, and he has a great time ... But ... you could get someone really mean two rows away from you and some people are really ... cruel. I'm not talking little kids who don't know what they're saying ... I mean adults.' (Klein et al., p. 593). As a consequence of this lack of consistency in people's behaviours and reactions, the usual sense of anonymity that other people experience when they are going about their normal lives is removed because the child with Apert syndrome stands out from the crowd and is visibly different. This could make every social experience unpredictable and children with Apert syndrome may receive very negative reactions from people just for being who they are. This has been very well explained by a mother who, when talking about the daily challenges her family faces, said '[You never know] how others will react. Going to a place that kids love—an amusement park—that should just be a happy time. But when you go it could be a happy time or a disaster, you never know. Things that people take for granted could always go in opposite ways.' (Klein et al., p. 596).

Mothers of children with Apert syndrome have highlighted the difficulties their children can face when they want to join in with sports activities with their peers (Klein et al., 2006). Because of their physical disabilities, some sports can be difficult. A mother of a boy with Apert syndrome reported that 'They're just faster than him, quicker. So it's hard ... They wanna do their thing and he can't keep up. So he usually stays with a lot of younger kids.' (Klein et al., p. 592).

Fearon and Podner (2013) highlighted the fact that many parents in their study reported that their children with Apert syndrome had particular difficulty with mathematics when compared to their reading and verbal skills. Parents also reported that many of their children had been diagnosed with attention deficit hyperactivity disorder.

Clearly, parenting a child with Apert syndrome is likely to provide challenges and experiences which are very different from those experienced by parents of typically developing children. These challenges and experiences are likely to have the consequence that the whole parent-child experience will be atypical, as parents try to provide their children with positive experiences and build their children's resilience, self-esteem and positive sense of self.

Conclusions

As can be seen, the development of children with Apert syndrome is the result of a complex interaction of their personalities, their physical abilities and disabilities and the world around them. These children face many challenges which may impact not only on the development of their cognitive and academic skills, but also their social, emotional and behavioural development. Existing studies on children with Apert syndrome are inconsistent, possibly due to the use of unreliable assessment data and the variability and complex needs of children with Apert syndrome. However, within this landscape of enormous unpredictability, there exists a need to try to begin to understand the challenges faced by children with Apert syndrome in order to help them engage socially, achieve academically and lead healthy and fulfilling lives.

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