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Neuropsychosocial Outcomes

of Patients With Untreated Craniosynostosis

ABSTRACT

Objective: To review research of visual, auditory and cognitive deficits alongside psychological impacts on patients with untreated craniosynostosis.

Methods: A 26-reference review from Pubmedallowed us to compile outcomes according to their experimental research.

Results: Auditory difficulties which have been identified, mainly in syndromic craniosynostosis were otitis media and deafness. Visual pathologies included exorbitism, astigmatism and strabismus, mostly in syndromic craniosynostosis. Cognitive deficits effect attention span, acedemic performance, memory and language. Lack of autonomy and inability to connect with others are few of the many psychological impacts that were found to correlate with craniosynostosis.

Conclusion: Future research should increase control groups of unaffected children and sampling sizes. This enables better comparison with more accurate mean values and thus any significant or insignificant deviances can be accounted for properly. Routine neuropsychological testing on such patients is recommended.

Keywords: Craniosynostosis, Complications, Untreated, Neuropsychosocial, Cognitive

1.0 INTRODUCTION

Craniosynostosis is the premature fusion of one or more sutures of the skull, namely metopic, lambdoid, sagittal and coronal sutures. It can present as an isolated deformity or as part of a syndrome. Pfeiffer, apert, crouzon, muenke and saethre-chotzen syndromes are the more common conditions that craniosynostosis manifests in.[1] Causes are mainly genetic, especially the syndromic forms, and the phenotype severity differs from mild to very severe. Namely, sagittal and metopic synostosis usually present with one clinical feature, a palpable bony ridge.[1] This creates a dilemma for parents in view of the risks associated with misdiagnosis as well as the risks of craniosynostosis being left untreated. If the parents or the patient never consent to treatment as the options may prove too difficult to decide upon, craniosynostosis may be left untreated. As a result, the raised intra-cranial pressure (ICP) leads to visual pathologies, auditory impairments and cognitive deficits.[2] All of these have a significant impact on thechild psychologically.

1.1 AUDITORY IMPAIRMENTS

Patients with craniosynostosis may have difficulty with audio, leading to disordered communication. Hearing loss mainly occurs in syndromic craniosynostosis, 91% of which is due to an FGFR-2 gene mutation.[3,4] As a result, speech is usually also impaired because word articulation proves difficult. This leads to a prolonged I-to-III interpeak latency of the auditory brainstem's response, which in turn form an abnormal wave II.[3] As shown in figure 1.1, anatomically, this pathology is probably due to raised ICP that compresses the auditory nerve as it is passing through the internal auditory meatus via the internal auditory canal.[3]

Associated complications included 27% that lost their hearing and all suffered from recurrent otitis media.[1,3,4] As shown in figure 1.2, this is recurrent inflammation of the middle ear treated by effusion. Assistive devices and sign-language can help such children to lead as much of a normal life as possible.

1.2 VISUAL PATHOLOGIES

The orbital area may be compromised as a result of craniosynostotic facial deformities which in turn lead to optic pathologies.

1.2.1 Exorbitism

The orbital area may be reduced due to the facial deformities present resulting in exorbitism. As shown in figure 1.3, the eyes of such patients protrude outwards; most commonly in patients with crouzon or apert syndrome.[1,7] Patients are more susceptible to corneal injury, infection or ulcers due to the upper eyelid being unable to protect the eyeball itself.[7]

1.2.2 Astigmatism and Strabismus

In syndromic craniosynostosis, the most commonly associated visual morbities are astigmatism and strabismus.[8] In isolated craniosynostosis, patientsare more susceptible to developing astigmatism in the eye contralateral of the fused suture.[1] Astigmatism is an abnormal curvature of the lens, leading to blurred vision as shown in figure 1.4. Patients present with a less spherical lens which results in an inability for light rays to meet at a common focus, thus forming a distorted image. Corrective surgery or lenses usually help these patients see clearer.[9]

The raised ICP may compress occular areas of the brain which in turn may lead to misaligned eyes known as strabsimus. As shown in figure 1.5, this varies according to the direction of misalignment. As a result, patients develop amblyopia (lazy eye) where the brain fails to process inputs from the effected eye and starts to favour the other. Vision weakens and tends to get worse if not aided by glasses, prisms, eye exercises or corrective eye muscle surgery.[11]

1.3 COGNITIVE DEFICITS

Older research has focused poorly on such cognitive outcomes as these lacked standard tests, control subjects and appropriate follow-up assesments. Currently, the Bayley Scales of Infant Development and the Wechsler Intelligent Scales of Children have looked upon mental deficits, not only in syndromic craniosynostosis but also in isolated forms.[1] Figure 1.6 explains how the patient's development is classified. Patients with single-suture craniosynostosis, such as unicoronal or unilambdoid synostosis, carry a

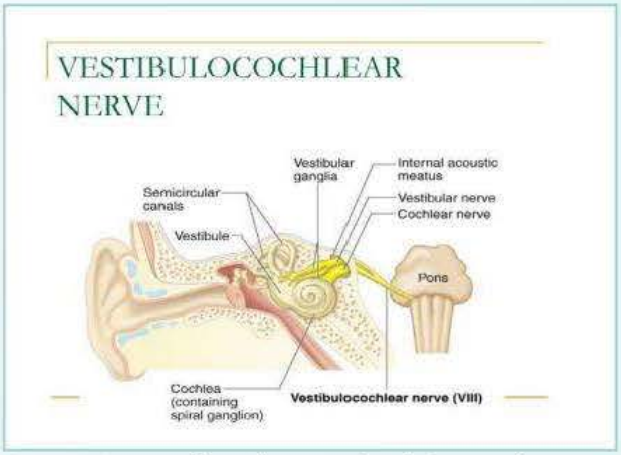

Figure 1.1: Passage of the auditory nerve (vestibular nerve & cochlear nerve)[5]

Figure 1.2: Normal Ear (left) and Acute Otitis Media (right)[6]

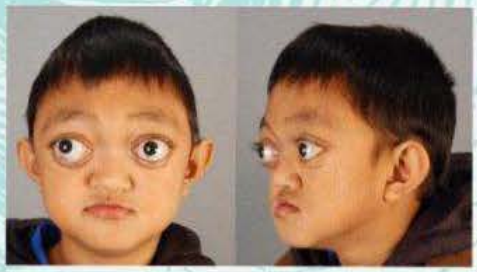

Figure 1.3: Seven year-old male with Crouzon Syndrome presents with exorbitism[8]

three to five-fold increased risk for such deficits.[13] Knight et al. proposed a systematic review in 2014 of 33 documents with special focus on methodology.[14] As a result, ten studies showed mental retardation, including language. Five studies of children averaging nine years old, showed normal IQ levels but three others showed increased learning disability and memory loss.[15,16] Cognitive imagery, attention span and acedemic performance were also recorded to be in deficit with patients of untreated craniosynostosis. Evidently, many assumptions with little research to back it up is present since many studies have contradicted each other.[17] However, in totality it can be concluded that some correlation between cognitive deficit and raised ICP exists.[17]

1.4 PSYCHOLOGICAL IMPACTS

The previous complications alongside the day-to-day challenges every person faces, leave a child with craniosynostosis conflicted and relatives in need of support in order to enable an easier life for their child. Along the stages of development, the family is confronted with ongoing medical decisions that need to be taken and psychological difficulties that need to be dealt with.[19]

1.4.1 Infant to School Age

During infanthood, the crucial psychosocial burden lies with the parents as they need to learn, adapt and manage their child's needs. Specifically in children with syndromic craniosynostosis, parents would have to deal with others' antipathetic reactions and be conflicted to choose their child's schooling. The child's appearance will also leave the parents confronted with dismissal from other parents thus, preventing child-to-child relationships.[1]

Once the child has begun to attend school, the challenges are posed more on the child rather than the parents alone. As a result, these children may find it difficult to make friends as they are dismissed. Furthermore, their reduced cognitive and motor skills may hinder them from participating in activites and academics, leaving these children at a disadvantage.

1.4.1.1 Medical Complications

Despite treatment, these children may still suffer from complications and unfortunately require hospitalisation. Approximately 10% of children admitted to the intensive care unit have experienced post-traumatic stress disorders (PTSD).[20] Parental stress and visibly reduced coping ability have been correlated strongly with PTSD in the child. Indicatively, the stress related to self-image, trauma, cognitive and motor deficits should not only be addressed and treated with the child, but also within the whole family.[1]

1.4.1.2 Abnormal Behaviour

The Child Behaviour Checklist has been used in a recent study by Becker et al. that reported significantly differing behaviour between craniosynostotic and uneffected people.[21] On the other hand, Vlugt et al. found no correlation as such when accounting for IQ.[22] Other studies taken during school age found that 33% of children with non-syndromic trigonocephaly, required a school psychologist. Approximately, 47% required remedial classes, 20% required special needs allowances and 37% were diagnosed with attention deficit disorder, autism and hyperactivity.[23]

1.4.2 Adolescense

In adolescense, approximately one third of patients have experienced bullying pertaining to stigma and appearance.[19] Most patients cope with continuous support and social interventions. Moreover, they are confronted with difficulty to be autonomous as they begin to reach adulthood and are required to consent and plan their own health-care with their specialists. It is emphasised that these patients should realistically form treatment expectations.[1]

1.4.3 Adulthood

Unfortunately, there have been little to no studies regarding non-syndromic craniosynostosis. Adults with Apert and Crouzon syndromes had a lower acedemic level, were less often married, experienced less sexual connections and had depressive mood periods.[24,25] However, they were just as likely to report a positive outlook on life in general. Indicatively, adults who experienced such depression were willing to undergo corrective surgery in adulthood despite the increased risks and complications.[26]

1.5 CONCLUSION

Indicatively, there are many ramifications that can occur if craniosynostosis patients are left untreated. In Malta, craniosynostosis is significantly rare with an average of two cases yearly from 1993 to 2016. The highest being 5 cases in 2011, all of which were live births. From a total of 28 cases in 24 years, only one fetal death in 1996 was reported. As a result, the total prevalance report as at 2016, was 2.71%.[27] The neuropsychosocial outcomes were highlighted in this review however, there are many others such as physiological and motor complications that were not mentioned

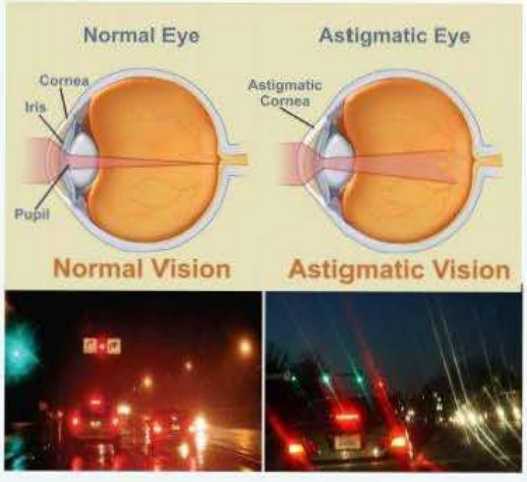

Figure 1.4: Normal vision meeting at a common focus (top left). Clear images during the night (bottom left). Astigmatic vision lacking a common focus (top right). Distorted images during the night (bottom right). Adapted from Health Jade[10]

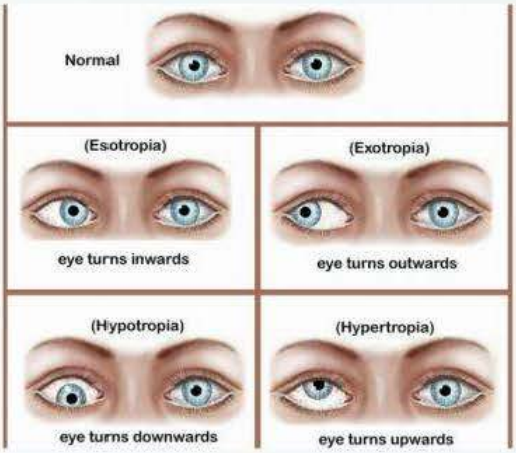

Figure 1.5: The different types of strabismus in comparison to the normal condition.[12]

in this review. It was deemed appropriate to focus on the cognitive and pyschological aspects as there is less insight on these topics as such. The visual and auditory impairments leave a child unable to communicate. The abnormal cognition leaves a child confused and struggling to make sense of things that others are thriving upon. All this with the added societal pressure and parental stresses leave a significant impact on the child's development, behaviour and his or her future relationships. This research has enabled a general review of all the possible long-term outcomes the patient may be confronted with. This emphasises the strong requirement of a well-explained set of facts to the patient so as to make the best decisions regarding their health care plan in a multi-disciplinary approach.

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Classification	Equivalent BSID-III Scaled Scores	Definition
At risk	1-4	Child is most likely in need of further evaluation to determine need for early intervention.
Emerging	5-7	Child has some risk for developmental delay, but further evaluation is made on the basis of other information collected. Can monitor develop or refer for further evaluation.
Competent	8-19	Child is considered at low risk for developmental delay and in most cases does not need further evaluation.

Abbreviation: BSID-III, Bayley Scales of Infant and Toddler Development, Third Edition.

Figure 1.6: Bayley Scales of Infant Development[18]

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