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Case Report

Holoprosencephaly: A Case Study for Communicative and Swallowing Management

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Introduction

Holoprosencephaly (HP) is a developmental central nervous system defect characterized by advanced facial anomalies resulting from the complete separation of two lobes of the cerebral hemispheres [1]. Prevalence is 1.31 in 10,000 births [2,3]. There is a parallel relationship between the combination of different etiological factors in different contributions and the changing clinical picture of the disease. Autosomal dominant and autosomal recessive inherited forms are reported in the literature [4,5]. Considering the studies on holoprosencephaly, developmental, neurological and medical problems were found in children with holoprosencephaly Cohen, 1982; Ersin & Ertugrul, 2005 [6,7]. Table 1 lists the physical findings that can be seen in individuals with HP. Postnatal care of HP is multidisciplinary, symptomatic and long-lasting cognitive and physical supportive therapy. Prognosis depends on the size of the associated medical and neurological disorders (Table 2).

Table 1: Physical characteristics that can be seen in individuals with HP.

Facial Anomalies (cyclopia, etmosephaly)
Facial midline defects (premaxillary agenesis)
Hyper-Hypo Telorism
Cleft palate / lip
Microcephaly
Straight or pointed nose bridge

Table 2: some accompanying problems of HP..

Neurocognitive deficits	Diabetus Mellutus
Epilepsy	Endoctrin anomalies
Recurrent Infections (Aspiration pneumonia)	Major organ malformations (Kardiac problems' kidney problems' etc)
Feeding problems	Otonomic nervous system problems (body temprature' heart rithm stability, etc)

There are HP and associated complications in the literature. However, in an HP case, language/speech impairment was not observed without dysphagia and craniofacial anomalies [8]. The purpose of this presentation is to share with you the therapeutic approach and results we have accomplished with this rare combination of severe neurocognitive-linguistic pathologies accompanied by severe dysphagia. We studied the case of a 24 month old girl with holoprosencephaly who was admitted to our hospital with difficulty swallowing. Mother was told about the possible problems prior to give birth as seen through ultrasound yet she and her husband chose not to terminate the pregnancy due to their religious beliefs (even though it was guaranteed by her medical primary care physician).

A multidisciplinary study of neurologist, nutritionist and dietician, language and speech therapist was provided during the diagnosis and screening period. Our case was born with a caesarean section at 38 weeks after completing the normal birth process and stayed in the cup for 28 days. It has been learned that she has had long-term jaundice after birth. The birth weight of our case was 2760gr and the neck was 50cm. It was seen that the values of life and weight were close to norms when compared with their peers. But the case was microcephaly and the head circumference was 32cm (n=35cm). There is no consanguineous marriage in the family history. The head control of the child was weak and could not sit without support. When you want to lift the foot, you have flexion and adduction in the hips, shearing in the legs, knee flexion,

and an equinovalgus posture in the ankle (Figure 1). It has been reported that the neurological examination is hypotonic and the emanation is weak, standing up. Cranial measurements are below normal. As shown in Figure 2(A-C), section of cranial MR images are compatible with semi lobar holoprosencephaly and have the features of:

A. Cerebral lobes are present but are fused anteriorly and at the thalami and then is partial diverticulation of the brain,

B. Absence of septum pellucidum,

C. Monoventricle with partially developed occipital and temporal horns,

D. Rudimentary falx cerebri: absent anteriorly,

E. Incompletely form interhemispheric fissure,

F. Fusion of thalami,

G. Agenesis of corpus collosum, and incomplete hippocampal formation.

Further, cavum septum pellucidum has not been observed, Lateral ventriculo-posterior horn width is 10mm and choroidal complex increases lateral wall distance. These results were also observed during fetal neurosonography that was also compatible with semi lobar holoprosencephaly. Posterior fossa was normal. At the time of clinical assessment, her eyes were able to follow visual and auditory stimuli. However, Denver Screening Test II Scores in combined with the AGTE (Ankara Developmental Test Inventory) indicated 3 months in general overall development age, 3 months in cognitive-linguistic skills development, 3 months in gross motor skills development in 3 months, and 3 months in social skill and self-care development 1 year and 2 months old. According to the findings of the anamnesis and the clinical evaluation, it was concluded that there was a "delay in cognitive development at a severe level". It was observed that there was an increase in tonus in the lower extremities. It has been also observed that some reflexes are still present even though they are diminished long before with age matched normally developing peers; DTRs live, moro +/+, clonus+/+, babinski +/+. Tongue-trusting was the primary means of bolus propulsion anterio-posteriorly, in that it takes more time and energy and less success with bolus to reach from oral phase to pharyngeal phase to swallow. Her teeth development was also irregular with enabling her to close for biting and chewing. Her palatal arch was also high which effected her palatal reflex to be triggered on a timely fashion.

Figure 1: Free stay of the child when lay on the bed.

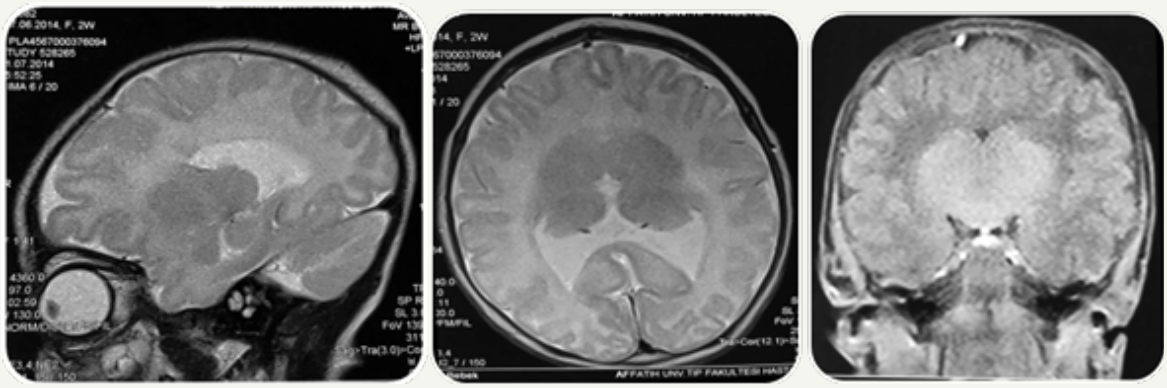

Figure 2: Cranial MR sections of sagittal (A), axial (B) and coronal (C) images.

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The clinic multidisciplinary team management included 6 weeks of therapy per our policy with severe problems as such and then to refer to related services (child life specialist, physical therapist, special education day care centre and dietitian) for continuity of the rehabilitation process. She was seen three times a week for 2, 5 hours/week to one-to-one therapy. Therapy focused on following directives, produce primary sounds that are toward age appropriate sounds (from vocalizations to start with), stay on focus and keep focus for 5-10 minutes, and decrease meal time with advancing bolus consistencies toward age appropriate level; such as, introducing different flavours other then milk, yoghurt and pudding. Implemented taping her face (Figure 3), thermal-tactile stimulation of facial pillars and neuro-electro stimulation of cricohyoid area as well as facial musculatures. Below table shows before and after status of 18 visits of intervention (Table 3,4). Knowing that it was not possible to make the differences to be normalized in this child, her family was very pleased with the changes in her over all improvement. They indicated that taking her out, playing with her socially and able to feed her without too much stress changed their quality of life (Table 5).

Table 3: Changes observed with therapy in the areas of speech/language.

Speech/Language	Before Therapy	After Therapy
First sounds	None (some weak vocals during laughing but not consistant	Vovels /ah, o, i and e/ with imitative cueing
Eye contact	Just for parents and some relatives that visits frequently	Clinicians and caregivers during therapy and medical care
Attention span	Not present	Able to focus and keep it for 10 minutes ongoing
Gesture development/use for communi-cation	Smile `happy face`	Smile, look for/search for, sorry/frawn (imitation of facial expressions)

Table 4: Swallowing.

Swallowing	Before Therapy	After Therapy
Oral-motor	Just palpational tongue movement, no lip closure	Started to close mouth, began to receivr food from the spoon and liquids fron a nosey cup
Bolus and saliva control	Very poor, drooling severely, has saliva burns around lower lip especially	Better saliva management, saliva burns lessened, bolus can stay in the mouth
Bolus formation	Not present, using syringe feed	Started to hold bolus in the mouth, less leakge with puree consistency
Meal completion time	150cc for 1,5 hour	150cc for 30 minutes
Meal consistency (solids)	Puding	Puree with some texture of soft mechanical consistencies
Liquid consistency	Pudding	Nectar

Table 5: Quality of life.

Family Perception	Before Therapy	After Therapy
Quality of Life Score	28	53
Beck Depression Score	Moderate level (score of 20)	Mild level (Score of 14)

To conclude, an intensive therapeutic intervention makes quality of life better for both family and the child's sake. It is a win-win situation for being able to do things more with their child since feeding time is considerately lessened, the family is learned how to interact with the child as seen in the Figure 2 (A-C) so that her vocalizations could be more productive turning into a communication style (sign plus some sounds and gestures in the future). Further therapeutic arrangements is also made that the family knows seating and positioning is need to be taken care of as well as the early child special education is a must for cognitive development.

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Figure 3: Taping of the facial muscles for lip closure and tongue mobility.

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