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Rare disease

The results of early physiotherapy on a child with incontinentia pigmenti with encephalocele

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Summary

This study aimed to exhibit the effects of early physiotherapy and discusses post-treatment results on a patient with incontinentia pigmenti (IP) with encephalocele. Physiotherapy evaluations of the child included cognitive, fine and gross motor development assessed with the *Bayley Scales of Infant and Toddler Development – Third Edition* (Bayley-III), disability level with the gross motor function classification system, gross motor function with the gross motor function measurement (GMFM), and tonus evaluation with the Modified Ashworth Scale. The child was included in a physiotherapy and rehabilitation programme based on neurodevelopmental treatment three times a week. Although cognitive and motor development according to Bayley-III improved in the present case, motor and cognitive retardation became more apparent with growth. GMFM results indicated a large improvement from 5.88% to 47.73%. Presentation of this case shows the significance of early physiotherapy in this first study on physiotherapy for IP during the early rehabilitation process.

BACKGROUND

Incontinentia pigmenti (IP) (Bloch–Sulzberger disease) was first diagnosed by Garrod in 1906.[1 2] IP is a rare genetic disease with characteristics of X-based dominant inheritance where dental, ocular, auricular, central nervous system (CNS), musculoskeletal and cardiovascular anomalies might coexist.[3–5] The cause is a mutation in the *NEMO* gene, which is a critical component involved in apoptosis and inflammatory responses and resides on chromosome Xq28.[6–9] The most apparent characteristics are skin symptoms observed as a result of non-holding of melanin in the epidermal basal cells and upper dermis melanophages.[4] Skin symptoms either diminish or totally resolve in adulthood.[10 11] Associated defects are dental abnormalities in 40%, central nerve system abnormalities in 30%, musculoskeletal and ocular abnormalities in 35%, and auricular or cardiovascular abnormalities and nail anomalies in 40% of patients. Immunological disturbances may also be observed.[3 12–15] Short height, short arms and legs, hemivertebra, hemiatrophy and kyphoscoliosis might be present as regards the musculoskeletal system.[16] Delay in the emergence of teeth, inadequate teething, cone-shaped teeth and dental abnormalities such as oligodontia can continue throughout life.[17 18] The CNS is the second most frequently affected system after the skin.[12] Motormental retardation (MR), epilepsy, microcephaly, spasticity, paralysis and ataxia conditioned by the CNS might be observed.[4 10] These symptoms are important since they are permanent, unlike the skin symptoms.[19–21]

Considering the course of the disease and its effects on various systems, the rehabilitation of IP necessitates a multidisciplinary team approach.[22] This team should include specialist doctors (paediatrists, paediatric neurologists, orthopaedists, neurosurgeons, neonatologists, child psychiatrists, dentists and all other related specialties), physiotherapists, child development specialists, dieticians, social workers and child care specialists. There is no prior study on the physiotherapy and rehabilitation of this disease. However, we think that it is necessary to consider children in the high-risk group within the context of early physiotherapy because of the symptoms and prognosis of the disease.

The early physiotherapy period includes approaches that begin with the neonatal period and continue until the 12th month.[23] Early physiotherapy and rehabilitation practices are preferred in disability situations conditioned by physical, mental, sense perception and cognitive problems caused by prenatal, natal or postnatal reasons.[23] It is reported that early physiotherapy can generally be effective thanks to brain plasticity and quick learning skills. A physiotherapy programme in this period enables normal movement facilitation, gaining functional movement, correct positioning and contact, early adaptation of the family to the disease, minimising future disturbances related to the musculoskeletal system and sensory perception/motor system deficiencies and effective determination of early period aims.[23]

The aim of this study was to demonstrate the effectiveness of early physiotherapy and discuss the post-treatment results of a child diagnosed as having IP.

CASE PRESENTATION

A 4-month-old child diagnosed as having IP was brought to the Cerebral Palsy Unit of the Hacettepe University Faculty of Health Sciences, Physiotherapy and Rehabilitation Department by her family, with the aim of receiving physiotherapy and rehabilitation. She had been born by spontaneous vaginal delivery at the 37th week of gestational age, with a birth weight of 2850 g. Her mother and father were third-degree relatives. She had cried and did not have any asphyxia or cyanosis at the moment of birth. Moro and sucking reflexes were observed to be normal and deep tendon reflexes were not present at the first neurological consultation at birth. Widespread oedema and wounds had been observed at birth and the mother and aunt also had similar histories themselves. Cranial ultrasonogram (USG)

for the oedema showed the existence of an 18×17 mm lesion extending to the sagittal interhemispheric fissure containing a solid cystic component. The results of an MRI performed in the first month included an arachnoid cyst in the posterior fossa, a porencephalic cyst in the left parietal lobe and diffuse contrast uptake of the tentorium. Electroencephalography (EEG) findings at rest and while awake were within normal limits.

TREATMENT

Physiotherapy evaluations at the early rehabilitation period

The following methods were used to evaluate the child at the 5th month (pretreatment), the 14th month (post-treatment) and during the bimonthly evaluations:

1. Demographic information of the patient (age, height, weight, head circumference; prenatal, natal and postnatal history): this was obtained from the medical records. Her height was 62 cm, weight 5500 g and head circumference 42 cm at the initial physical consultation.
2. *Bayley Scales of Infant and Toddler Development - Third Edition* (Bayley-III): the Bayley-III is a revision of the *Bayley Scales of Infant Development – Second Edition* (BSID-II).[24] The Bayley-III is a norm-referenced developmental scale over the first 42 months that has good psychometric properties and assesses infant and toddler development across five domains: cognitive, language, motor, social–emotional and adaptive.[25] It is also used in other childhood disorders such as cerebral palsy.[26] We used the cognitive and motor scales of Bayley-III. The composite score was interpreted as follows: 130 and above, very superior; 120–129, superior; 110–119, high average; 90–109, average; 80–89, low average; 70–79, borderline; 69 and below, extremely low.[25]
3. Disability level: the disability level of our case was determined using the Turkish gross motor function classification system (GMFCS).[27]
4. Gross motor function: the gross motor function of our case was measured using the gross motor function measurement 88 (GMFM-88).[28]
5. Tonus evaluation: the presence, absence and intensity of any spasticity were evaluated using the Modified Ashworth Scale (MAS).[29]

Physiotherapy programme

A physiotherapy programme based on Bobath neurodevelopmental treatment (NDT) principles was applied three times a week. A home programme and home education were also provided for the family.

Our aims in the early physiotherapy programme based on NDT principles were positioning, normal postural control and facilitation of movement reactions with particular handling positions, and improvement of functional motor development while providing symmetrical middle line orientation, increasing normal movement of extremities, increasing sensory input including proprioception and tactile stimulation[23 30]

The home programme and family education included providing information, adaptation of handling techniques into daily life and positioning that will increase the child's communication with the environment and make it easier to obtain nutrition and support, normal motor development, independent sitting and weight balancing, and teaching the family the techniques of handling that are appropriate for the treatment aims. The family was checked on a weekly basis to determine whether they had applied the home programme correctly.[23 30]

OUTCOME AND FOLLOW-UP

The Bayley scale of infant and toddler development

The Bayley-III change in composite scores of the patient is presented in table 1. Cognitive and motor development improved during the rehabilitation process in our case.

Level of disability

The level of disability of our case according to the GMFCS was level V at the fifth month and level III at the end of the treatment programme. GMFCS defines level V as the level at which physical impairments limit voluntary control of movement. Infants are unable to maintain antigravity head and trunk postures while prone and sitting and require adult assistance to roll. A child at level III can maintain floor sitting when the low back is supported and can roll and creep forward on his/her stomach.

Gross motor function

The gross motor function of the child showed marked improvement between pretreatment and post-treatment values (figure 1). The GMFM-88 scores (%) were 6.21%, 11.66%, 16.66%, 26.42% and 47.73% at months 5, 7, 9, 12 and 14, respectively.

Tonus evaluation

Our patient's tonus was found to be normal at all assessments.

Table 1 Motor and cognitive composite *Bayley Scales of Infant and Toddler Development – Third Edition* (Bayley-III) scores

Date	Motor			Cognitive		
	Composite score	Percentile	95% CI	Composite score	Percentile	95% CI
5 Months	70	2	65 to 80	55	0.1	51 to 67
7 Months	76	5	70 to 86	65	1	60 to 76
9 Months	61	0.5	56 to 72	60	0.4	56 to 72
12 Months	58	0.3	54 to 69	60	0.4	56 to 72
14 Months	64	1	59 to 75	65	1	60 to 76

DISCUSSION

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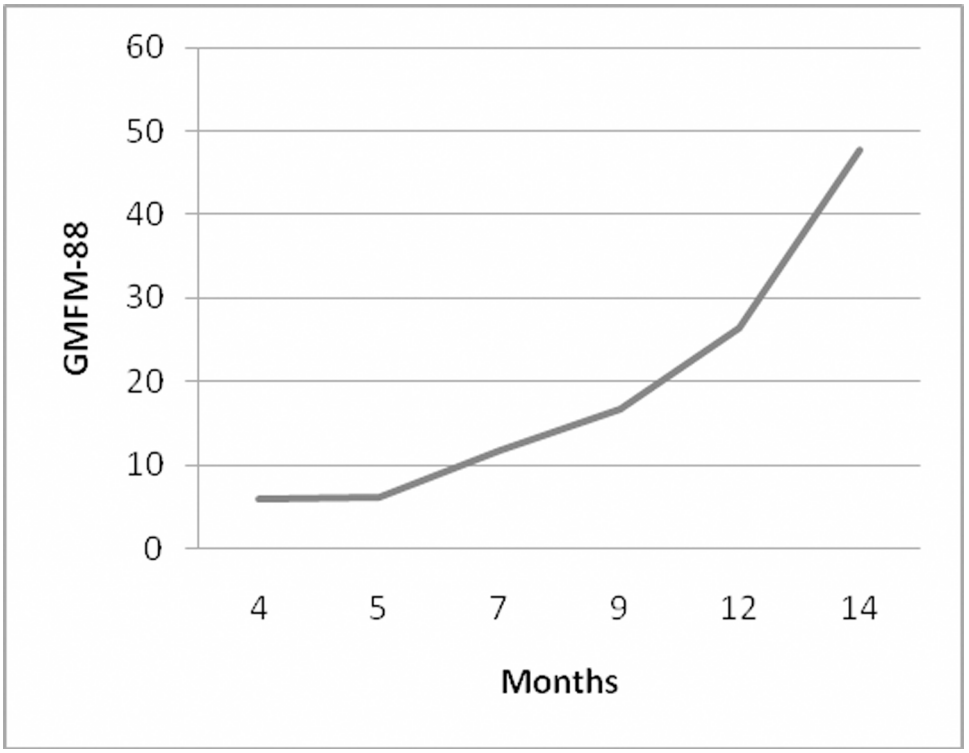

Figure 1 Gross motor function measurement (GMFM) scores of the patient.

IP is mostly seen in infant girls in the postnatal period as a result of an X chromosome mutation that might be accompanied by anomalies in other organs and systems, and has a frequency of occurrence of 1/40 000.[9 16] Our case was a 4-month-old girl.

Anomalies in other systems are observed frequently in IP disease. Ocular anomalies are observed in 35% of the cases and include widespread retinal pigmentation, microphthalmia, lenticular haemorrhage, retrolental fibroplasia, cataract and optic atrophy,[17 31] and our case showed ocular signs consistent with the literature as well. An eye examination when the child was 10 months old showed a normal optic nerve but there was difficulty moving the left eye upward and downward. Our observational evaluations showed that the patient had no ability to follow objects.

CNS dysfunction is observed in 30% of patients with IP.[16] Neurological symptoms are reported in the first week following birth and visualisation of brain damage by MRI is performed at the third day at the earliest.[32 33] Microcephaly, stroke, MR, spasticity and ataxia may be present. MRI anomalies include heterotopias, hemimegalencephaly, focal cortical dysplasia, callosal digenesis, cortical atrophy and cortical malformations such as periventricular lesions.[12] Spinabifida, anencephalia and encephalopathy are widespread neural tube defects in IP.[17] Our case had corpus callosum agenesis, CNS abnormalities, Dandy–Walker syndrome, an arachnoid cyst the in posterior fossa, a porencephalic cyst in the left parietal lobe as a posterior and compatible lesion, diffuse contrast abnormalities and an encephalocele.

Surgery for the encephalocele and duraplasty was performed on our case when she was 9 months old. It is generally reported that the visual and neurological prognoses are good in children with IP if ocular and CNS anomalies are not observed in the first year.[31] However, there were anomalies in both systems in our case.

General studies in the literature related to IP cases mostly focus on the medical condition or the systemic abnormalities, and studies on early physiotherapy results were not available until recently. The most commonly observed neurological disability in IP cases is MR, which was present in our case.[33 34] MR is classified as mild in children whose IQ is 50–70 and severe when the IQ is below 50. Beckung *et al*[35] investigated motor and emotional dysfunction based on the child's level of MR and epilepsy. The motor and emotional developmental level of none of the children involved in the study was compatible with their age. None of the 57 children with severe MR had normal gross motor function, fine motor function, coordination, balance or perception. However, 7 children out of the 31 with mild MR had normal gross motor function but 2 had epilepsy that was difficult to control. Normal participation was observed in six children, one of whom had uncontrollable epilepsy and five had controlled epilepsy. The GMFM gross motor function score was 65% in children with CP with mild MR and 22% in those with severe MR,[35] showing that MR severity affected the child's motor function and performance. Forssberg states that limited system function and sensorimotor afferents give the child very little stimulus and this results in weak body image and activity paucity. Children with MR show lack of

motivation that mostly affects undevelopedmotor behaviour.[36] We believe that the MR in our case was a negative factor affecting the physiotherapy process and activity.

Beckung *et al*[35] state that individual and group physiotherapy can help a child if gross and fine motor function, coordination, balance and participation is present. While children with CP and MR need physiotherapy on a regular basis, children who only have MR and epilepsy still need to be evaluated by a physiotherapist and supportive advice provided to the families to support their child's normal development even if treatment is not required on a regular basis.[35] The first evaluations performed in our case showed the baby's need for a regular physiotherapy programme and the necessity of supporting it with a home programme and family education.

The BSID-II is the current 'gold standard' for assessing a child's general development from 1 month to 42 months of age.[37 38] Bayley-III is used for the evaluation of the child's development and the effectiveness of therapeutic approaches in many paediatric disabilities[26] and we therefore chose to use Bayley-III for our case with IP. We did not have a chance to compare our case with another child with the same diagnosis, as no other studies on physiotherapy in children with IP are present in the literature. Cognitive and motor development improved in our case during the rehabilitation process, but we believe that the motor and cognitive retardation became more apparent with growth.

It is emphasised in the literature that early physiotherapy approaches are practiced for children with a risk of neuromotor dysfunction to minimise the level of possible future disability.[39] Yigit *et al*[39] have studied the benefits of the early physiotherapy approach on premature babies. Their survey research population included 229 babies who were born before the 34th week and weighed less than 2000 g. They could not demonstrate the effect of early physiotherapy on motor functions of premature children with low birth weight but did show the importance of a routine CP development risk monitoring programme.[39] A successful treatment has been reported in 93% of the 27 studies on an early rehabilitation programme but the majority of these studies are based on subjective data and observations. The authors have found statistical evidence that supports a benefit in 48% of the studies.[40]

The Ottenbacher *et al*[41] study showed that the motor development levels of children receiving NDT were more advanced than children being treated otherwise. We did not have the chance to compare our case with others, as this is the first article on the physiotherapy and rehabilitation practices of children with IP. However, we performed comparisons with our patient's evaluation results over time and also with other high-risk infants with different diagnoses reported in the literature.

Learning points

- We observed that the physiotherapy programme has to be started immediately after birth to support the developmental process in our case with incontinentia pigmenti (IP) that we followed-up during the early rehabilitation process.
- This first presentation of an IP case that underwent physiotherapy during the early rehabilitation process has demonstrated the importance of early physiotherapy.
- The next step should be long-term studies on a larger number of cases with a multidisciplinary team.

Competing interests None.

Patient consent Obtained.

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