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Though Early Cardiac Involvement Can Be Seen With Novel Mutations

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Abstract:

We evaluated a Comorian girl aged 3 years 6 months with neuroregression and seizures. Te child was third born to second-degree consanguineous parents by lower-segment cesarean section due to cephalopelvic disproportion. Birth weight was 3.1 kg with uneventful perinatal history. Tere was maternal history of normal healthy live birth in the frst pregnancy and spontaneous miscarriages in second and fourth pregnancies in early trimesters. Te fourth pregnancy was associated with Down syndrome. Te mother was given antenatal progesterone for excess bleeding in the frst trimester.

Introduction

Terefore, reconstruction of dysfunctional fexor tendons is occasionally necessary following compartment syndrome. In this report, we introduce the technique of early palmaris longus (PL) tendon transfer for FPL reconstruction and describe the surgical outcomes in a patient with compartment syndrome treated using early PL tendon transfer

By revealing genetic defects in anaplastic kinase (ALK)-positive non-small cell lymphoma Lung Cancer (NSCLC), High Potency ALK Tyrosine Kinase Inhibitors (ALK-TKIs) are developed for therapy.

obstacle to achieving a high level of influence in the centernervous system (CNS) [1]. Because of its effectiveness, lorlatinib is a common treatment option for patientsALK-positive non-small cell lung cancer with one or more ALK-TKIs No success.

A 57-year-old Japanese man complained of tinnitus and was presented to the otolaryngology department of our hospital. A contrast-enhanced computed tomography (CT) scan showed a tumor in the upper lobe of the left lung, approximately 40 mm in the long axis.

The hilar and mediastinal lymph nodes were also enlarged. Suspected primary lung cancer, the patient was referred to the Department of Otolaryngology and Respiratory Medicine on the day of admission comfirmed.

Te results of the 18F-FDG PET/CT and other fndings led to the diagnosis of lung adenocarcinoma [cT2bN2M1c (PUL, OSS); stage IVB]. We recommended rebiopsy considering the possibility of a genetic mutation, but the patient refused the examination because of strong coughing during the frst bronchoscopy and progressive symptoms of hoarseness. Te patient strongly desired an early treatment, so we initiated frstline therapy with cisplatin (75 mg/m2) and pemetrexed (500 mg/m2) 3 weeks after the frst visit.

U pacienta sa však vyvinula renálna dysfunkcia 2. stupňa [kreatinín, 1,83 mg/dl; odhadovaná rýchlosť glomerulárnej filtrácie (eGFR) a vidíme, že ide o čiastočnú odpoveď, ale opacity skla a infiltrácia sa naďalej šírili oboma smermi.V tomto čase sa pacient sťažoval na aktívnu dýchavičnosť a perkutánnu saturáciu kyslíkom. je asi 82 % vo vnútornom ovzduší. Analýza plynov ukazuje, že parciálny tlak kyslíka je 58,2 torr, čo naznačuje zlyhanie dýchania. Testy funkcie pľúc ukázali obštrukčné poškodenie dýchacích ciest s vitálnou kapacitou (VC) 2,11 l a %VC 57,1 %.

Discussion and conclusions

In the search for advanced lung cancer treatment, driver genes have been identifed, and several molecular-targeted drugs have been developed and reported to exhibit remarkable antitumor efects. For ALK-positive NSCLC, alectinib was more efective than crizotinib in phase III trials (ALEX study) and is, therefore, widely used for frst-line treatment. Lorlatinib and ceritinib are efective against alectinib-resistant lung cancer and have been used in second-

line treatments, based on the status of the resistance gene. Lorlatinib, which is derived from crizotinib, is structurally optimized to inhibit mutations conferring resistance, including G1202R, and those with a high rate of migration to the CNS. In the CROWN trial, the intracranial response among patients with measurable brain metastases at baseline was 82%, with a complete intracranial response rate of 71%. In the global ALEX, ALTA-1L, and eXalt3 trials, the corresponding complete intracranial response rates with alectinib, brigatinib, and ensartinib were 38%, 28%, and 27%, respectively. Moreover, in the CROWN trial, lorlatinib signifcantly decreased the cumulative incidence of CNS progression, which suggests that the prolonged progression-free survival seen with lorlatinib may be partly due to the prevention of CNS metastases. In the CROWN trial, progression-free survival was signifcantly longer among patients with ALK-positive NSCLC who received frst-line lorlatinib than among those who received crizotinib. In untreated patients, lorlatinib may eliminate rare preexisting subclones harboring ALK resistance mutations or prevent the emergence of such resistant subclones .

We evaluated a Comorian girl aged 3 years 6 months with neuroregression and seizures. Te child was third born to second-degree consanguineous parents by lower-segment cesarean section due to cephalopelvic disproportion. Birth weight was 3.1 kg with uneventful perinatal history. Tere was maternal history of normal healthy live birth in the frst pregnancy and spontaneous miscarriages in second and fourth pregnancies in early trimesters. Te fourth pregnancy was associated with Down syndrome. Te mother was given antenatal progesterone for excess bleeding in the frst trimester.

History and physical examinations pointed toward the diagnosis of GM2 gangliosidosis (Tay–Sachs disease, SD, AB variant). In view of cherry-red spots and coarse facies, GM1 gangliosidosis was also considered. No signifcant abnormality was noted in complete blood count, electrolytes, or renal and liver function tests. Ultrasonography of abdomen did not reveal any hydronephrosis or other anatomic abnormalities. Computerized tomography scan of brain without contrast was suggestive of mild bilateral symmetric hyperdensity of thalami (Fig. 3). Electroencephalogram (EEG) showed slowing of delta frequencies associated with drowsiness. Video-fuoroscopic assessment for swallowing function was suggestive of aspiration on both fuoroscopic runs. Magnetic gaining weight till 5 months of age, after which there was fattening of the growth curve and failure to thrive. At 6 months, she had developmental arrest followed by progressive neuroregression. She also had severe startle response since 8 months of age. Ten, she started having generalized recurrent seizures from 9 months onward. Te epileptic episodes were mostly focal with secondary generalization, with the most severe event reported as having frequency of ten seizure episodes within 2 hours time period despite anticonvulsant therapy. She had also macrocephaly with coarse facial features, persistent laryngomalacia, and hyperacusis. Tere was no muscle atrophy. Central hypotonia, peripheral hypertonia, and a positive Babinski refex were elicited. Organomegaly was absent. Ophthalmological examination showed bilateral macular cherry-red spots and an inability to fxate the eyes. At 12 months, she developed gastroesophageal refux disease (GERD) as well as reactive airway disease. Gastrostomy tube feeding was also commenced. She had frequent episodes of hospitalizations due to repeated aspiration pneumonia, reactive airway diseases, and other central nervous system complications. History and physical examinations pointed .

Te wound was closed with vessel tape, utilizing the shoelace technique. After the release, the patient was not able to fex the interphalangeal joint of the thumb. At second-look operation (3 days after the initial release), swelling of the left forearm had reduced with no evidence of infection. Terefore, we performed open reduction and internal fxation for the left radius fracture and PL tendon transfer to the FPL tendon. Intraoperatively, the presence of the intact PL tendon was confrmed. Te PL tendon was advanced to the FPL tendon, and an interlacing suture was performed thrice. Te tendon was transferred with the wrist in 30° of dorsifexion and the metacarpophalangeal and interphalangeal joints of the thumb in 45° of fexion.

Early intervention with tendon transfer is usually not warranted in areas with open wounds; nevertheless, early PL transfer soon after injury is a simple procedure that does not require an additional surgical approach. Even if the transferred PL tendon does not regain a sufcient amount of function, other tendons can be used as salvage donors, with minimal impairment of hand function. We emphasize that early tendon transfer with PL can provide satisfactory results, and could be considered as a potential choice for FPL reconstruction following compartment syndrome of the forearm.

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