We present 2 cases of hearing loss (HL) with developmental delay due to Cockayne syndrome (CS) which is characterized by normal prenatal growth with the onset of growth and developmental abnormalities in the first two years. Both of them developed HL prior to diagnosis of CS. There HL were both progressive. CS is one of the cause of hearing and vision impairment.

Introduction
Cockayne syndrome is a rare autosomal recessive disorder caused by a primary defect in DNA repair. A diagnostic feature of CS is the impaired recovery of RNA synthesis in fibroblasts of patients after UV irradiation. Clinically it is characterized by neonatal growth retardation, impaired development of the nervous system, hearing loss, premature aging, cachexia, and characteristic facies and multiple ocular problems. The clinical spectrum of CS encompasses a wide range of severity. Patients have been divided into subgroups based on age at disease onset as well as on symptom severity: type II patients are characterized by the presentation of canonical symptoms from birth and by a short life span (2–7 years), type I patients display most symptoms in their first 2 years of life and have a mean life expectancy of 16 years, and type III patients show a later onset of the disease and have a prolonged survival until adulthood.

Case 1
The patient was a 4 years old boy at the time of the first visit for hearing examination. He had mild developmental delay with unknown cause. Audiometric evaluation revealed mild HL (Fig.1a). His hearing level was gradually deteriorated, and the threshold of Auditory brainstem response (ABR) was 50dBnHL at the age of 5 (Fig.1b). He started to wear hearing aids. His language comprehension and expression were significantly delayed.

He was also diagnosed as pigmentary degeneration of the retina around the same time. He also had characteristic face. Therefore, CS type I was suspected, and clinically diagnosed at the age of 5 with short stature, impaired neurological development and calcification in brain CT.

His hearing level was 65dB at the age of 6, and it was 95 dB at the age of 14 (Fig.2). ABR revealed that thresholds for wave V were 90 dBnHL in right ear and 80 dBnHL in left ear at 12 years.

He wore hearing aids and glasses. His visual impairment was also progressive. His hearing was more useful than his vision in his later years.

Case 1
The patient was a 4 years old girl at the time of first visit for our department. She had been treated for otitis media with effusion for a year. She also had mild developmental delay with abnormal findings in brain MRI. At the age of 4, she was diagnosed moderate HL of unknown etiology (Fig.3a). The threshold of ABR was 50dBnHL on both ears. She started to wear hearing aids, and it was effective.

She was diagnosed as pigmentary degeneration of the retina at the age of 7, and later she was suspected CS. CS was confirmed by a specific DNA repair assay on fibroblasts from skin biopsy. Any gene mutation hasn’t been detected in CSA nor CBA gene. Her HL was progressive, the threshold of ABR was 70dBnHL in right ear and 90dBnHL in left ear.

She is 8 years old, her hearing and vision loss is progressive (Fig.3c).