The pena-shoker syndrome type i : clinicopathological report of a recurrent case.

Pena-Shokeir syndrome is a rare lethal disorder wich clinical phenotype is the result of a deformation sequence caused by fetal akinesia/hypokinesia [1] . In approximately 50 % of cases an autosomal recessive mode of inheritance were documented [2]. we report a case of a Pena – Shokeir syndrome type I with positive family history.

CASE REPORT

Mrs B.K, 39 years old, married since 5 years, no consanguinity and mother of two childrens. In her first pregnancy she doesn't underwent any medical control. At term, a female was born with polymalformatif syndrome (multiple ankylosis, pulmonary hypoplasia and facial dysmorphism), dead after some hours with severe breathing difficulties. In the second and the third pregnancy she had two normal female babies. In actual pregnancy, no medical control was observed. She presented at 32 weeks a premature labour. Clinical examination noted polyhydramnios with abdominal distension. The foetal cardiac rythym was normal. Echography shows a singleton in cephalic position with hypokinesia, multiple arthrogryposis, lung hypoplasia and hypertelorism. No other abnormality was seen. Biologic check up and karyotype were normal. Foetal IRM shows cerebellar atrophy with vermix cerebelli agenesia (Fig.1, Fig.2). Interruption of pregnancy was decided. Newborn was wheiting 1850 g, presenting multiple arthrogryposis, camptodactyly, facial dysmorphism and ogival palate (Fig.3). She dead 20 minutes later. Postnatal autopsy confirmed the prenatal findings.

Figure 1 : Foetal IRM: cerebellar atrophy with vermix cerebelli agenesia.

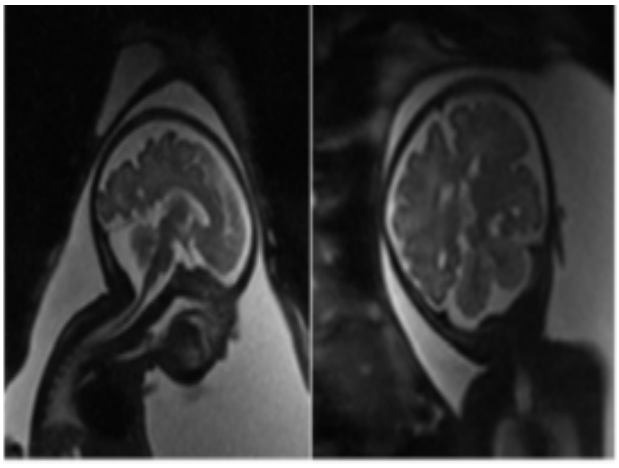


Figure 2 : Foetal IRM : multiple arthrogryposis, lung hypoplasia

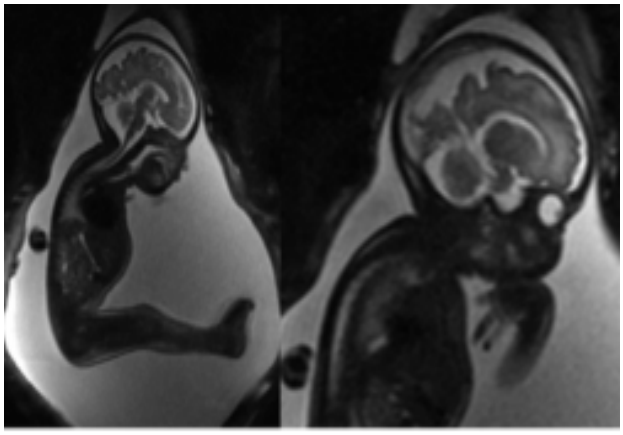
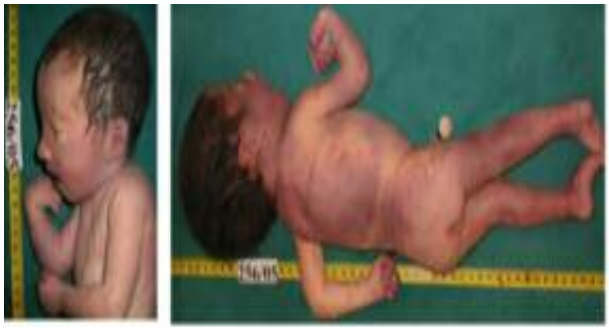


Figure 3 : arthrogryposis, camptodactyly, facial dysmorphism



CONCLUSION

Prenatal sonography and foetal IRM play an important role in detecting Pena – Shokeir syndrome and in providing the detailed pattern of fetal akinesia. Postnatal recognition requires genetic counselling of parents and obtaining early prenatal diagnosis in next pregnancy.

REFERENCES

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