A Case of an Unusual Presentation of Faltering Growth

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Abstract

Five weeks old infant presented with poor feeding, failure to thrive and excessive sweating, apart from being underweight her all systemic examination was unremarkable except tachycardia. Because of tachycardia and part of work up an echocardiography was requested, which showed hypertrophic cardiomyopathy. She later developed hypertension and was treated accordingly.

A left renal mass was picked on ultrasound of abdomen, so urine catecholamine levels were done which were elevated confirming the diagnosis of neuroblastoma. Cytogenesis of the tumour showed favourable histology and was classified as infantile neuroblastoma and fell in the intermediate risk group. Chemotherapy was started followed by surgical resection of the tumour and she was gradually weaned off her medication. It was suggested that the hypertrophic cardiomyopathy was due to the excessive release of catecholamine. She has been doing very well since then.

Keywords: Faltering Growth; Poor Feeding; Cytogenesis

Introduction and Case Report

We would like to report a case of an unusual presentation of faltering growth in five weeks old infant.

Five weeks old infant presented with poor feeding, failure to thrive and excessive sweating, apart from being underweight her all systemic examination was unremarkable except tachycardia. Because of tachycardia and part of work up an echocardiography was requested, which showed hypertrophic cardiomyopathy. She later developed hypertension and was treated accordingly.

A left renal mass was picked on ultrasound of abdomen, so urine catecholamine levels were done which were elevated confirming the diagnosis of neuroblastoma. Cytogenesis of the tumour showed favourable histology and was classified as infantile neuroblastoma and fell in the intermediate risk group. Chemotherapy was started followed by surgical resection of the tumour and she was gradually weaned off her medication. It was suggested that the hypertrophic cardiomyopathy was due to the excessive release of catecholamine. She has been doing very well since then.

Neuroblastoma

Neuroblastoma is the most common malignant solid tumour of the peripheral nervous system in infants and young children [1]. It accounts for 8 - 10% of all childhood tumours and causes 15% of all cancer deaths in children [2]. 90% of the cases are diagnosed under 5 years, 30% of which present within the first year of life [3]. It is rare in adolescents and carries a poor prognosis in this age group. Males are more commonly affected (1.2:1) [4].
Neuroblastoma is associated with many genetic abnormalities including DNA content, gain of chromosome arms 17q or deletion of chromosome arms 1p and 11q [5]. The clinical presentation depends upon tumour location, size, degree of invasion, effects from catecholamine release and presence of paraneoplastic syndrome. Most common site is abdomen (65%), half of these are localized in adrenal medulla. The other locations could be neck, chest or pelvis, in 1% primary tumour is not detected [4,6]. Patients can be asymptomatic or can have constitutional symptoms like fever, malaise, weight loss; mass, pain, abdominal distension, lymphadenopathy, constipation or urinary retention in pelvic tumours, dysphagia, dyspnoea in thoracic tumours [7] and Horner's syndrome in cervical tumours. Epidural extension of the tumour causes progressive paralysis in 15% of the patients [4].

At initial diagnosis 50% patients will have localized disease while 35% can have lymph node metastasis. It usually metastasizes to bone marrow, liver and bone. Proptosis and periorbital swelling (raccoon eyes) is seen with orbital involvement while blue subcutaneous nodules (blueberry muffin syndrome) are seen with skin dissemination [2,4]. The excess of catecholamine's cause hypertension and tachycardia. Paraneoplastic syndrome can present as intractable diarrhoea (due to excessive release of VIP) and opsoclonus-myoclonus syndrome [2].

Neuroblastoma is divided based on the degree of neuroblastic differentiation into undifferentiated, poorly differentiated and differentiating and on the basis of mitosis-karyorrhexis index into low, intermediate or high [8]. International neuroblastoma risk group staging system classifies neuroblastoma into L1 (localized disease), L2 (localized disease with image defined risk), M (distant metastasis) and MS (metastasis to skin, liver and/or bone marrow in children < 18 months). Based on this patients are divided into very low, low, intermediate and high risk group. According to international neuroblastoma staging system NB is classified into stages 1 - 4 with a special stage 4s which occurs in infants < 1 year [1].

The diagnostic tests include basic blood work chest x-ray, urine and serum catecholamine and their metabolites. Elevated levels of LDH, ferritin and neuron-specific enolase are associated with advanced stage or relapse [9]. Imaging includes CT scan of neck, chest, and abdomen and MRI spine. Other tests include bilateral bone marrow biopsy, MIBG for detection of primary tumour and metastasis, FDG-PET for soft tissue metastasis and FISH for MYCN amplification and other chromosomal aberration [2,10]. Diagnosis is confirmed by histology on specimen obtained from tumour [2].

The treatment is dependent on the stage and risk stratification. It consists of chemotherapy, surgical resection and radiotherapy. Induction chemotherapy reduces the tumour mass allowing for complete surgical resection followed by consolidation phase. Surgical resection is not recommended in infants with stage 4s as these tumours regress spontaneously. Radiotherapy is needed in high risk patients [2].

Prognosis depends upon age at diagnosis, site of primary tumour, histology, lymph node involvement, response to treatment and biological features. 5 year survival rate has increased for all age groups but specifically for infants < 1 year [10].

Conclusion

Neuroblastoma can present in different ways, in our case it presented with faltering growth, hypertension and hypertrophic cardiomyopathy.

Bibliography


