Case Report

Stridor in a neonate - Is it just a floppy larynx?

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Abstract

22q II deletion syndrome, characterized by deletion of long arm of chromosome 22, encompasses a wide range of clinical features, mainly congenital heart defects, facial dysmorphism, palatal defects, feeding problems, immune deficiency and hypocalcaemia. We report a case of 8 days old baby with 4 day history of stridor, feeding problems and vomiting. He was found to have some dysmorphic features and proven to have this deletion syndrome on FISH (Fluorescent In Situ Hybridization) testing. He was then effectively managed by a multi-disciplinary team effort. Clinicians shoud have low threshold of karyotyping and FISH in a neonate presenting with stridor along with dysmorphic features.

Introduction

About 1 in 200 babies are born with a chromosomal abnormality.[1] Some of these children are phenotypically normal, while others have obvious, sometimes, severe manifestations of disease.[1] These disorders usually present with a varied combination of cardiac, facial, palatal and neurological features. Even in this modern era, they present a management challenge for the paediatricians worldwide. We report a case who was diagnosed to have a chromosomal disorder based on a high index of suspicion and then managed by a multi-disciplinary team effort.

Case Report

We report a case of an infant who presented when 8 days old with a 4 day history of stridor, feeding problems and vomiting. According to the mother, the stridor was worse during feeding and was progressively increasing in intensity. On examination, he was noted to have a few dysmorphic features like small, receding chin, prominent nose and bilateral fixed talipes (Figure 1). He was also noticed to be desaturating markedly while feeding; thereby started on intermittent oxygen therapy. Meanwhile, blood was sent for karyotyping and in view of progressively worsening stridor and increasing oxygen requirements, he was seen by the ENT team who performed an endoscopy and confirmed mild laryngomalacia. Moreover, in view of significant gastro-oesophageal reflux, treatment with anti-reflux medication was commenced.

Subsequently, results from the Cytogenetics Laboratory reported deletion of long arm of chromosome 22, confirmed by FISH (Fluorescent In Situ Hybridization) (Figures 2). These findings led to a diagnosis of 22q11 deletion syndrome.

We then performed further investigations to look for other features of this deletion syndrome. Apart from a slightly reduced CD4 cell count, rest of the tests including serum calcium, echocardiograph, ultrasound renal tract and hearing test were all normal.

We arranged a multi-disciplinary team meeting for appropriate management of the case prior to discharge from

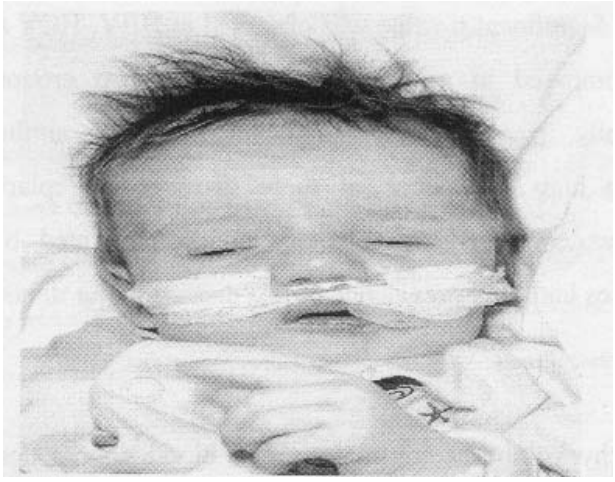

Figure 1 Patient's Photograph revealing dysmorphic features (small, receding chin, prominent nose).

Figure 2. Karyotyping photograph (arrow indicating deletion of long arm of chromosome 22).

the hospital. Genetic counselling was done by the Consultant

Paediatricians and subsequently by the Geneticist; we also sent blood from both parents for karyotyping . His feeding problems were dealt with by the dietitian in coordination with the nursing staff. His bilateral fixed talipes was managed by Orthopaedics with fortnightly renewal of his bilateral leg plasters. Similarly, laryngomalacia was being looked after by ENT team. He was continued on anti-reflux medications. A recent 24 hours single channel pH study was normal. He has been referred to the Community Paediatrician for his ongoing care in the community. Social services input was required for support at home and the family is in receipt of appropriate disability living allowances.

The baby was finally discharged home on air, stabilised on feeding. The parents were given resuscitation training before discharge. Follow-up was arranged with Consultant Paediatrician in order to co-ordinate overall care, ENT team for laryngomalacia and Orthopaedics for talipes. We plan to arrange further immunological studies, once our patient has received all his vaccinations (no live vaccines have been given to our patient).

Discussion

22q11 deletion syndrome is characterized by a sub-microscopic deletion of chromosome 22 detected by Fluorescent In Situ Hybridisation (FISH). It is now recognized that this syndrome encompasses the phenotypes previously called DiGeorge syndrome, Velocardiofacial syndrome, Conotruncal anomaly face syndrome, many cases of the autosomal dominant Opitz G/BBB syndrome and Cayler cardiofacial syndrome. Estimates of prevalence vary from 1 in 4000 to 1 in 6395.[2-3] Major clinical features include congenital heart defects (74%)[4], characteristic facial features[5], palatal abnormalities (69%)[6], learning difficulties (70-90%)[7], immune deficiency (77%), hypocalcaemia (50%) and feeding problems (30%).[8]

Our case presented with stridor, which is not a common presentation of this syndrome. The only positive findings included bilateral talipes (antenatally detected), small chin, prominent nose and poor feeding. There was no evidence of cardiac or palatal abnormalities or low calcium levels. Still, having a high index of suspicion, we sent the blood for karyotyping which came back positive for 22 q 11 deletion.

As we found out later, diagnosing this condition was just a first step in managing this complex disorder. We arranged a multi-disciplinary meeting to cover all the aspects of management. Participants of this meeting included supervising Paediatrician, ENT Consultant , dietetian, speech and language therapist, physiotherapist, social worker, general practitioner, community and hospital based nursing staff and the parents.

Important aspects of management included discharge plan, genetic counselling of parents, care of feeding and follow up with Orthopaedic and ENT teams.

Therefore, our case illustrates the importance of keeping a low threshold for suspicion of chromosomal disorders in newborn and managing these disorders with a competent multi-disciplinary team.

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