

Case Report

Facial palsy: a rare manifestation in Fanconi anaemia

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ABSTRACT

Fanconi anemia (FA) is an autosomal recessive genetic disorder characterized by progressive pancytopenia, multiple congenital anomalies, increased susceptibility to acute myelogenous leukemia and epithelial cancers especially in head and neck and GUT. The otologic manifestations in patients with Fanconi anemia is only about 10%. The present case report highlights the rare manifestation of Facial palsy/otologic and other associated anomalies in a 5 year old boy with Fanconi anemia.

Keywords: Fanconi anemia, Facial palsy, Congenital abnormalities, Ear anomalies, Aplastic anemia

INTRODUCTION

Fanconi anemia (FA) was first described by the Swiss pediatrician Guido Fanconi in 1927 is an autosomal recessive condition and clinically presents as a heterogenous disorder characterized by congenital malformation, pancytopenia and predisposition to malignancy. Skeletal abnormalities are most common anomalies, followed by skin pigmentation, and short stature whereas central nervous system defects are rare anomalies.¹ Auricular malformations, conductive hearing loss and external auditory canal stenosis are the most common otologic presentations but are usually not present in the majority of cases.² Here, we present a case report to highlight the rare manifestation of facial palsy/otologic and other anomalies in Fanconi anemia.

CASE REPORT

A 5-year-old boy provisionally diagnosed a case of Fanconi anemia was referred to ENT department with chief complaint of epistaxis, gum bleeding and ear malformation. Patient reported with recurrent fever, cough, pallor and nasal bleeding and was admitted in pediatric department. Patient had short stature, mild

microcephaly, microphthalmia, pallor, bilateral absent thumbs and presence of supernumerary thumb as shown in Figure 1, hypo/hyper pigmented patches over skin. There were low set ears, right pinna was malformed, microtia on left side with absent cartilage as shown in Figure 2, bilateral very narrow meatus, tortuous external auditory canal, tympanic membrane partly visualized, and tuning fork tests showed moderate conductive hearing loss. Patient was not cooperative for audiologic evaluation.



Figure 1: Bilateral absent thumbs



Figure 2: There were low set ears, right pinna was malformed, microtia on left side with absent cartilage.

Tongue was pigmented/geographic tongue. Most importantly patient had facial palsy on left side with deviation of angle of mouth to right and incomplete closure of left eye as shown in Figure 3. Ultrasonography showed horseshoe shaped kidney and chromosome breakage study showed positive results.



Figure 3: Facial palsy-left side with incomplete eye closure.

DISCUSSION

Fanconi anemia presents with birth incidence of 1 per 350000. Most cases of FA manifest anaemia symptoms during childhood.³ It is associated with progressive bone marrow failure, congenital anomalies, growth retardation, hyperpigmentation of the skin, café au lait spots and a predisposition to malignancies, especially acute myelocytic leukemia and squamous cell carcinoma.⁴ About 75% of patients with Fanconi anemia have birth defects, such as altered skin pigmentation and/or café au lait spots (>50%), short stature (50%), thumb or thumb and radial anomalies (40%), abnormal male gonads (30%), microcephaly (25%), eye anomalies (20%), structural renal defects (20%), low birth weight (10%), developmental delay (10%), abnormal ears or hearing (10%).⁵ In the present case, patient presented with short stature, hypo/hyper pigmented patches over skin, mild microcephaly, microphthalmia, pallor, bilateral absent thumbs and presence of supernumerary thumb. There were low set ears, right pinna was malformed, microtia on left side with absent cartilage, bilateral very narrow meatus, tortuous external auditory canal and tympanic membrane was partly visualized.

The most common audiologic manifestation in Fanconi anaemia is asymmetrical bilateral conductive hearing loss that was more severe at lower frequencies and in some cases had a progressive character.⁶ In our case there was moderate conductive hearing loss on tuning fork test. The other anomalies are developmental delay, Bell's palsy, CNS arterial malformation, stenosis of the internal carotid.⁵ In the present case, left facial palsy is noted with deviation of angle of mouth & incomplete closure of eye. This feature of facial palsy in FA has not much been documented in past cases. Facial palsy is very rare in Fanconi's anaemia and was noted in present case.

The differential diagnosis of FA also includes hereditary bone marrow disorders with radial ray defects, such as thrombocytopenia-absent-radius syndrome, Diamond-Blackfan anemia, conditions with increased chromosomal breakage, such as Nijmegen breakage syndrome, and conditions associated with aplastic anemia, such as dyskeratosis congenita and Shwachman-Diamond syndrome.⁴

Fanconi anemia can be accurately diagnosed using diepoxybutane (DEB) induced chromosome breakage which is highly specific. The cells of patients with FA are characterized by chromosomal hypersensitivity to cross linking agents and the resulting increase in chromosome breakage provides the basis for a diagnostic test.⁷ In the present case, chromosome breakage study was positive.

If a diagnosis of FA is suspected, it should be confirmed by chromosome breakage. Genetic counselling and cytogenetic testing of the patient's family should be done. All patients should have an ultrasound of the renal tract, hearing tests, and a haematological assessment that should include examination of the bone marrow. HLA typing in anticipation of possible bone marrow transplantation should be performed. An endocrinological assessment should also be made.⁷

Androgens are used to boost haematopoiesis. Colony stimulating factors can be used in conjunction with androgens or in place of them if they have failed. Blood transfusions are used to maintain counts and treat symptomatic problems. Bone marrow/haematopoietic stem cell transplantation using related donors (where possible) is the only curative treatment. After transplantation, careful monitoring for evidence of leukaemia/solid tumours is necessary.⁸

In general bone marrow transplantation should be performed at a young patient age, before the use of androgens and with normal platelet levels at the initiation of the transplant, has been associated with a good survival.⁴ Androgens, especially oxymetholone to increase bone marrow cellularity. The latter drug is associated with increased risk of acne, hyperactivity, premature epiphyseal closure and hepatic tumours.⁷

Thereafter, gradually decreasing the androgen dose at 2-4 month intervals. If, in the absence of other causes of cytopenias (such as viral or bacterial infection), no response is seen after 3-4 months, oxymetholone should be discontinued.⁹

The main aspect associated with the use of corticosteroids in acute phase of Bell's palsy is that inflammation and edema of the facial nerve are implicated in causing Bell's palsy and corticosteroids have a potent anti-inflammatory action which should minimise nerve damage and thereby improve the outcome.¹⁰

In the past literature, randomized, double-blind, placebo-controlled trials have provided compelling evidence that treatment with prednisolone improves outcome in patients with Bell's palsy and shortens the time to complete recovery.¹¹

Extensive chemoradiation used in the transplantation procedure can be highly toxic because of the underlying DNA repair defect in FA. Histocompatible (matched) sibling donor transplant remains the best treatment for FA and gives the best outcome, if performed early.¹² Still, transplant survivors experience multiple complications, including physical injury from chemoradiation (including pulmonary and renal toxicity and veno-occlusive disease), graft-versus host disease (GvHD), immune injury, sterility, and endocrinopathies.¹³

The prognosis of this genetically heterogeneous disorder is poor, with 35% of patients dying at a median age of 13 years due to the frequent respiratory infections caused by leucopenia. Prognosis is good with children treated with bone marrow transplantation.¹⁴

CONCLUSION

Fanconi anemia is a clinically heterogeneous disorder. Conductive hearing loss, external auditory canal stenosis, and auricular malformations are the most common otologic presentations. In this case report, we present Fanconi's anaemia with ENT manifestations which are least present in majority of cases. Facial palsy is very rare manifestation of FA and was noted in the present case. This feature of facial palsy in FA has not much been documented.

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