JOHANSSON BLIZZARD SYNDROME

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Abstract:
Johansson Blizzard syndrome is an extremely rare autosomal recessive multi-system congenital disorder featuring abnormal development of pancreas, nose and scalp with mental retardation, hearing loss and growth failure sometimes described as a form of ectodermal dysplasia. Here we are reporting a female neonate with the characteristic features.

CASE REPORT
A baby girl was delivered normally with a birth weight of 1.8 kg. There was no parental consanguinity. On birth the most striking feature was hypoplastic alae nasi giving a beak like appearance of her nose, frontal upsweep of hair, prominent eyes with mongloid slant, long philtrum, thin upper lip, micrognathia, low set ears, microcephaly, single palmar crease and anteriorly placed anus.

Baby was started on breast feeding and started having diarrhoea. Thyroid function test revealed low T3, low T4 and normal TSH levels. Her blood indices, skull x-rays and chest x-ray and ultrasonogram abdomen revealed no abnormality. Date was fixed for brainstem evoked response audiometry in the department of ENT for assessment of hearing. The diagnosis of Johansson Blizzard syndrome was made based on the characteristic facial features, hypothyroidism, pancreatic insufficiency and growth retardation noted in the child.

DISCUSSION:
Johansson Blizzard syndrome was named after Drs. A.J. Johansson and Robert M.Blizzard

Keyword: Johansson Blizzard syndrome, hypoplastic alae nasi, hypothyroidism, autosomal recessive
Johanson Blizard syndrome
Johansson Blizzard syndrome is a rare autosomal recessive multisystem disorder. The most prominent feature is exocrine insufficiency of pancreas (100%). Craniofacial anomalies include mild to moderate microcephaly (50%), midline scalp defect most typically posterior, but can be anterior or over vertex (87%), variable sparse hair with frontal upsweeping (96%), hypoplastic to aplastic alae nasi (100%), nasolacrimal duct fistulae (66%), hypoplastic deciduous teeth, absent permanent teeth (90%). Anorectal anomalies include imperforate anus or anteriorly placed anus (40%), rectoureteral or rectovaginal fistulae (18%).

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Genitourinary anomalies include caliectasis to hydronephrosis, vagina septae, micropenis, hypospadias or single urogenital orifice. Also hypothyroidism of unknown etiology (30%), mental retardation sometimes severe (67%), sensorineural deafness (75%), hypotonia (80%) and prenatal onset of growth deficiency (60%).

The molecular basis of JBS has recently been mapped to chromosome 15q15-q21 with identified mutation in the UBR1 gene. UBR1 encodes one of the several E3 ubiquitin ligases of the N-end rule pathway, an ubiquitin dependent proteolytic pathway. Ubiquitylation and subsequent degradation of proteins at the proteasome is the universal mechanism for regulated protein degradation and the control of many intracellular protein levels.

While there is no cure for JBS, treatment and management of specific symptoms and features of the disorder are applied. Pancreatic insufficiency and malabsorption can be managed with pancreatic enzyme replacement therapy. Craniofacial and skeletal abnormalities may require surgical correction using techniques including bone grafts and osteotomy procedures. Sensorineural hearing loss can be managed with the use of hearing aids.

BIBLIOGRAPHY: