General Discussion

Johanson-Blizzard Syndrome (JBS) is an extremely rare inherited disorder characterized by an unusually small nose that appears "beak shaped" due to absence (aplasia) or underdevelopment (hypoplasia) of the nostrils (nasal alae); abnormally small, malformed primary (deciduous) teeth and misshapen or absent secondary (permanent) teeth; and/or unusually sparse, dry, coarse scalp hair that tends to have a distinctive "upsweep" in the forehead area. In addition, affected infants may have a low birth weight, demonstrate signs of insufficient intestinal absorption (malabsorption) of fats and other nutrients due to abnormal development of the pancreas (exocrine pancreatic insufficiency), and fail to grow and gain weight at the expected rate (failure to thrive) during the first years of life, contributing to short stature.

Approximately one third of infants with Johanson-Blizzard Syndrome also demonstrate abnormally decreased activity of the thyroid gland and underproduction of thyroid hormones (hypothyroidism), causing generalised weakness and contributing to growth retardation, as well as abnormal delays in the acquisition of skills requiring the coordination of mental and physical activity (psychomotor retardation). In many cases, affected infants may also exhibit hearing impairment of both ears at birth due to abnormalities of the inner ear (congenital bilateral sensorineural hearing loss) and may experience associated, severe speech impairment. In addition, approximately 60 percent of affected children have moderate mental retardation; however, others may have normal intelligence or mild retardation. In many cases, additional abnormalities may also be present. The range and severity of symptoms may vary greatly from case to case. Johanson-Blizzard Syndrome has autosomal recessive inheritance.