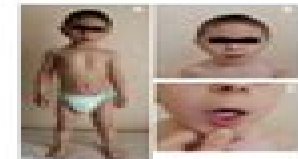
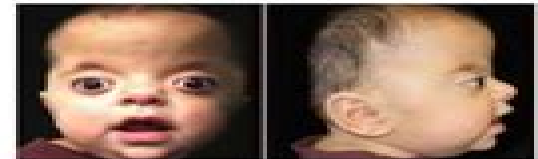


- Diagnosis- family history of condition, DNA marker tests, foetal ultrasonography, clinical evaluation, CT scans, MRI scans
- Treatment- depends on the symptom. Braces, dental surgery,
- **Apert Syndrome**- distinct malformation of the skull, face, hands and feet and the fibrous joints between the skull will fuse too early and this will lead to a pointed head
 - Signs and Symptoms- premature closure of the skull joint but brain is still growing and the pressure of the growth causes lots of bones to change their shape and can form a pointed head which is known as acrocephaly, wide spaced eyes, bulging eyes, cleft palate, flattened nose with low bridge, open bite, short fingers broad thumbs, partial/ complete fusion of the fingers and the feet
 - Causes- mutations in the FGFR2 gene and this plays a critical role in skeletal development
 - Diagnosis- clinical evaluation of the physical features, congenital heart defects may be noticed, MRI, hearing impairments using the new born hearing test and testing the FGFR2 gene
 - Treatment- supportive therapies, surgery to correct the bones, shunt to drain excess CSF, surgery for the hands and toes, early intervention is important
- **Grieg Syndrome**- deformities of the digits and the craniofacial area
 - Symptoms- extra fingers and toes, webbing and fusion of the fingers and toes, large skull, high, prominent forehead, broad nasal bridge, permanently flexed fingers, wide spaced eyes
 - Causes- autosomal dominant and abnormal variant of the GLI3 gene
 - Diagnosis- thorough clinical evaluation, X rays, CT scans, head circumference for macrocephaly
 - Treatment- craniofacial reconstructive surgery but not as common as widely spaced eyes and a big head are not enough for this, surgery for the toes
- **Foetal Valproate Syndrome** baby is exposed to valproic acid or sodium valproate in the first three months of the pregnancy. Valproate is needed for seizures
 - Symptoms- history of valproate use, spina bifida, vertical fold of skin on either side of the nose, underdeveloped nails of fingers and toes, hip dislocation, absence of the first rib, microcephaly
 - Causes- exposure to valproate
 - Diagnosis- history of valproate use, exclusion diagnosis
 - Treatment- avoiding valproate in pregnancy but if it cannot be avoided, administer it as a single drug, advice on taking folate acid
 - Treatment- speech therapy behavioural therapy, spina bifida surgery, correcting the heart defects using surgery
- **Trisomy 21**- also called Down's syndrome which is cost by the trisomy of the chromosome 21
 - Symptoms- physical growth delays, intellectual disabilities, characteristic facial features, low IQ, stunted growth, low muscle tone, flat head, deviated first toe, large tongue, brushfield spots
 - Diagnosis- screening for Down's physical examination and seeing the chromosome 21, ultrasound
 - Management- education, good environment, screening for the associated problems, proper care.
- **Trisomy 18**- also called Edward's Primary syndrome which is a trisomy of chromosome 18
 - Symptoms- kidney malformations, intestines protruding out of the body, microcephaly, small jaw, cleft lip and cleft palate, dropping the upper eyelid, CNS abnormalities
 - Diagnosis- ultrasound, PAPP-A and free hCG levels
- **Trisomy 13**- chromosome 13 is a triploid chromosome and it can be a mosaic presentation and the severity of the conditions will depend on the location of the chromosome 13 gene and whether or not it is mosaic



Edward's Syndrome



Review Of Human Development

CO Houle



Review Of Human Development:

The book delves into Review Of Human Development. Review Of Human Development is a vital topic that needs to be grasped by everyone, from students and scholars to the general public. The book will furnish comprehensive and in-depth insights into Review Of Human Development, encompassing both the fundamentals and more intricate discussions.

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