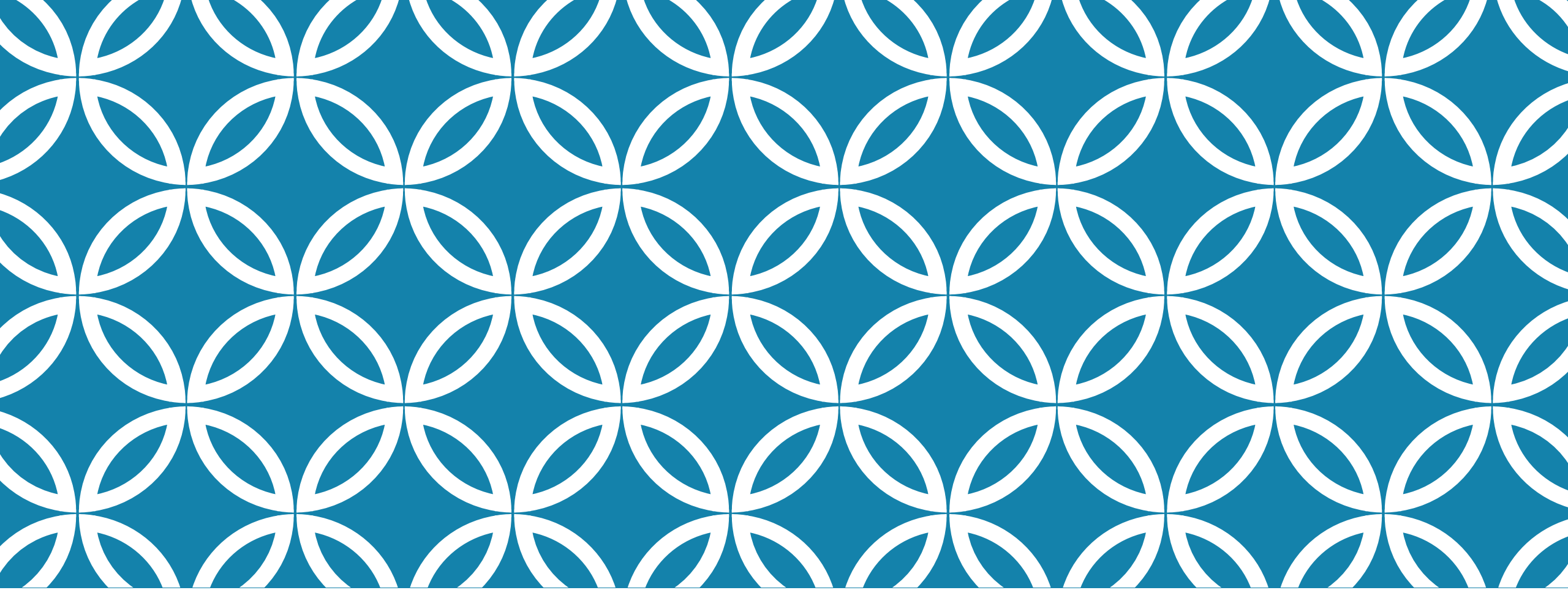


HEREDITARY DISEASE | Cri du chat

GROUP

Aashish kumar patel

Nitish

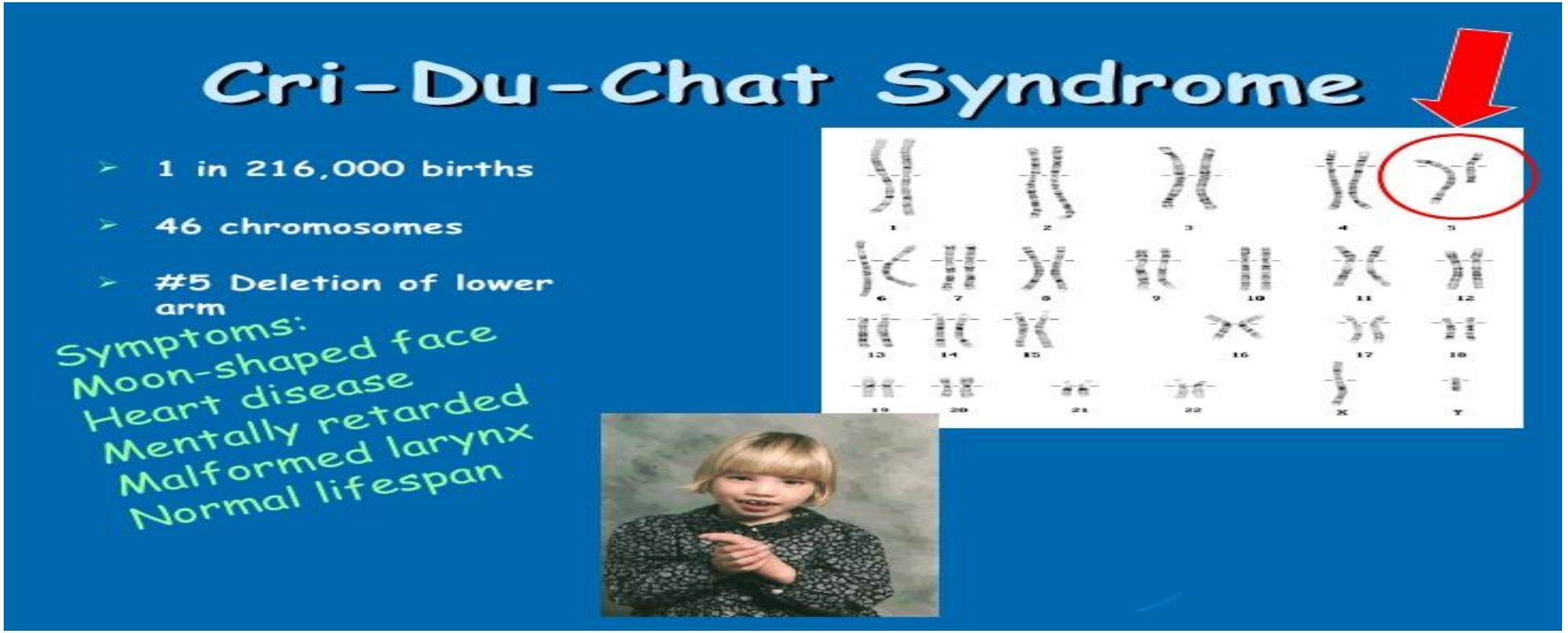
Cri du chat syndrome - also known as 5p- syndrome and cat cry syndrome - is a **rare genetic condition** that is caused by the deletion (a missing piece) of genetic material on the small arm (the p arm) of chromosome 5. The cause of this rare chromosomal deletion is unknown

Signs and symptoms

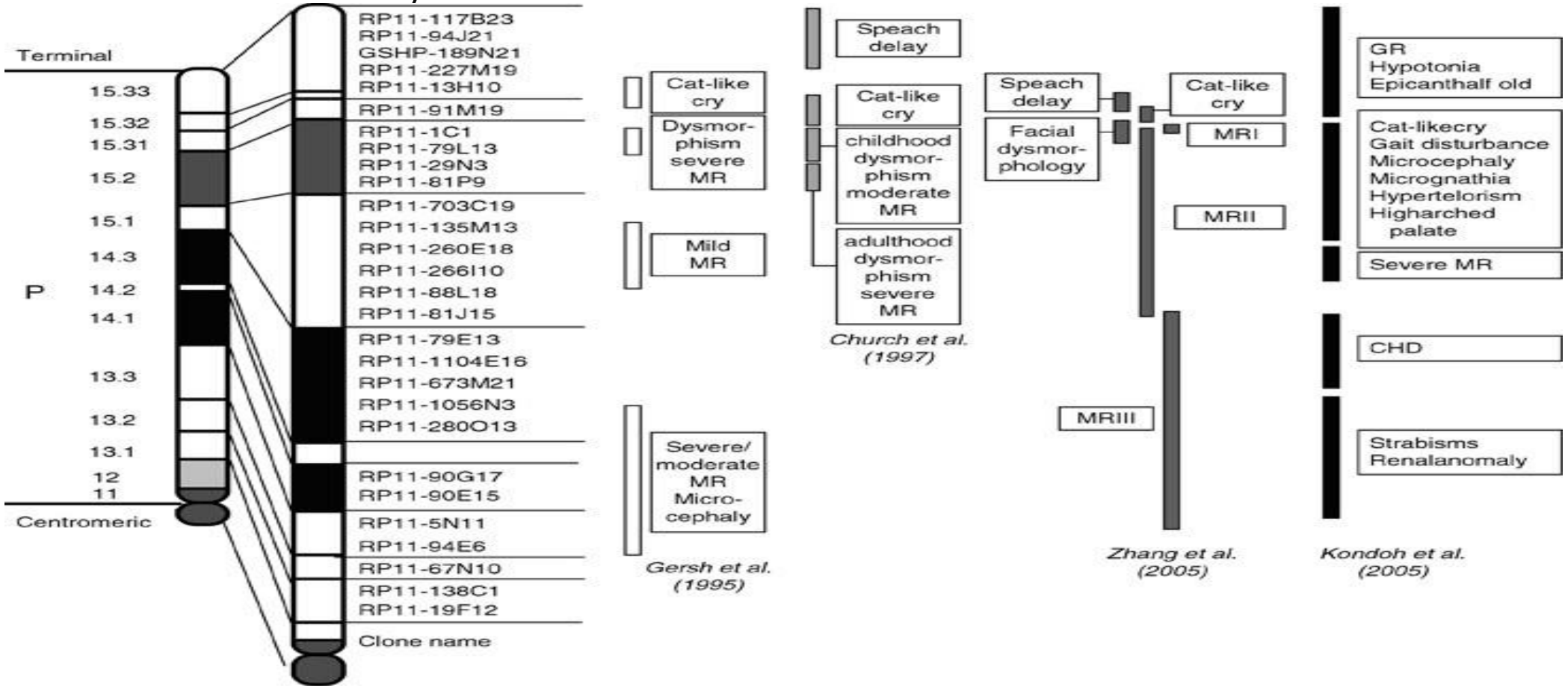
The syndrome gets its name from the characteristic cry of affected infants, which is similar to that of a meowing kitten, due to problems with the larynx and nervous system. About one third of children lose the cry by age of 2 years. Other symptoms of cri du chat syndrome may include:

- feeding problems because of difficulty in swallowing and sucking;
- mutism;
- low birth weight and poor growth;
- severe cognitive, speech and motor disabilities;
- behavioural problems such as hyperactivity, aggression, outbursts and repetitive movements;
- unusual facial features, which may change over time;
- excessive drooling;
- small head (microcephaly) and jaw (micrognathism);
- widely-spaced eyes (hypertelorism);
- skin tags in front of eyes.

OTHER COMMON FINDINGS INCLUDE HYPOTONIA, A ROUND FACE WITH FULL CHEEKS, EPICANTHAL FOLDS, DOWN-SLANTING PALPEBRAL FISSURES (EYELIDS), STRABISMUS, FLAT NASAL BRIDGE, DOWN-TURNED MOUTH, LOW-SET EARS, SHORT FINGERS, SINGLE PALMAR CREASES AND CARDIAC DEFECTS (E.G., VENTRICULAR SEPTAL DEFECT [VSD], ATRIAL SEPTAL DEFECT [ASD], PATENT DUCTUS ARTERIOSUS [PDA], TETRALOGY OF FALLOT). INFERTILITY IS NOT ASSOCIATED WITH CRI DU CHAT.

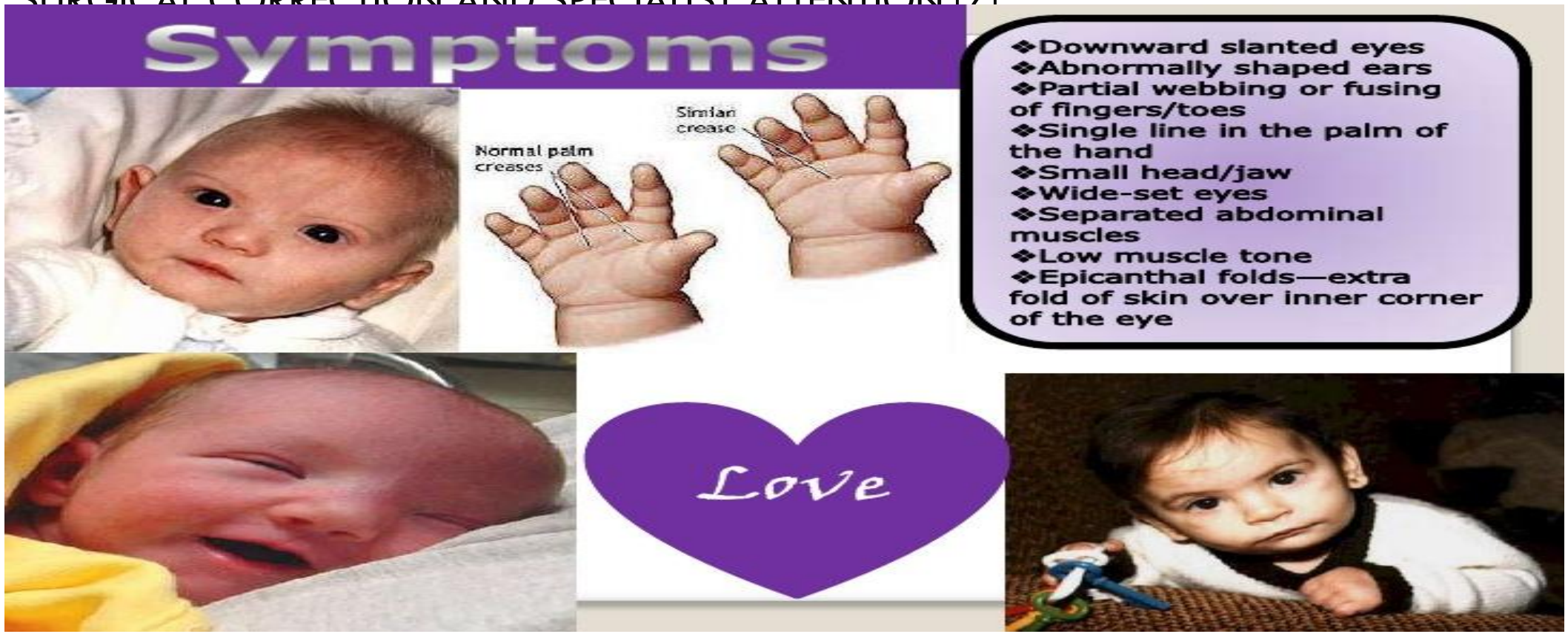


IT HAS ALSO BEEN OBSERVED THAT PEOPLE WITH THE CONDITION HAVE DIFFICULTIES COMMUNICATING. WHILE LEVELS OF PROFICIENCY CAN RANGE FROM A FEW WORDS TO SHORT SENTENCES, IT IS OFTEN RECOMMENDED BY MEDICAL PROFESSIONALS FOR THE CHILD TO UNDERGO SOME SORT OF SPEECH THERAPY/AID WITH THE HELP OF A PROFESSIONAL.

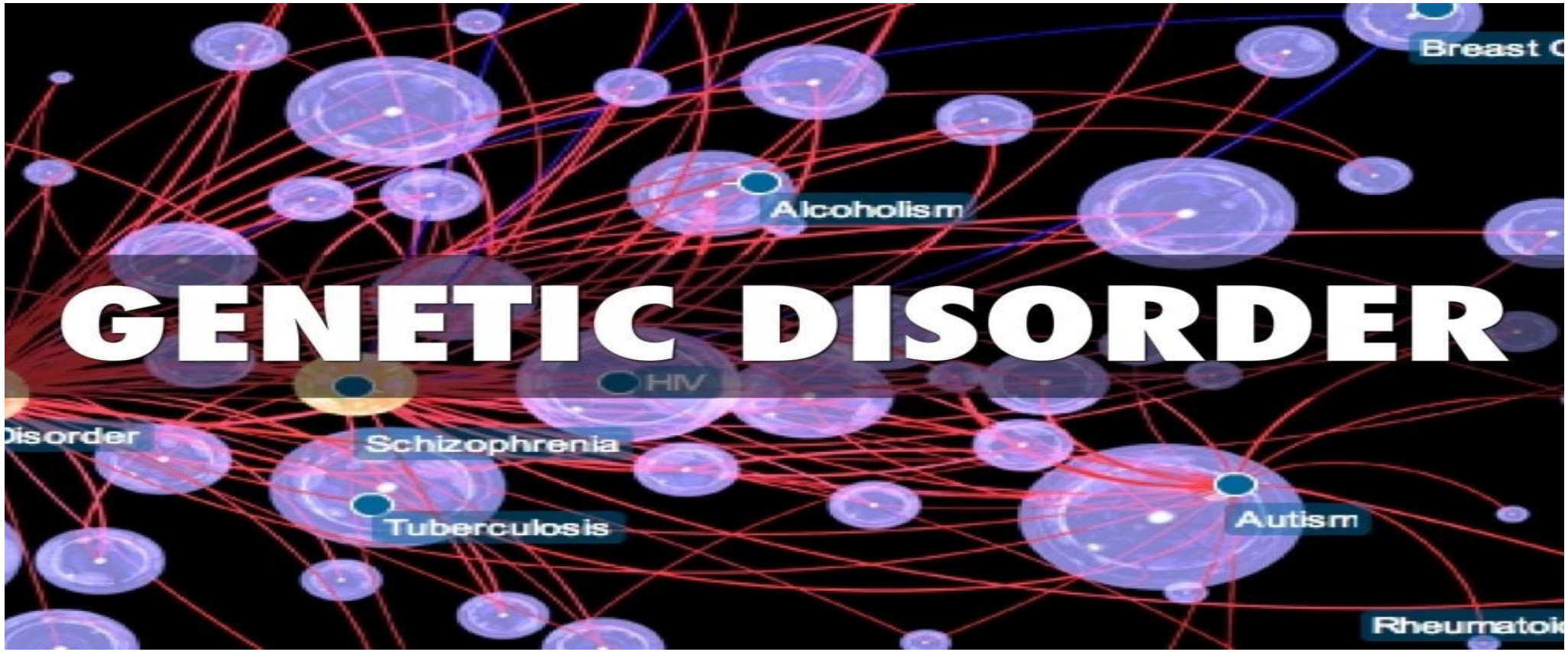


CRI DU CHAT SYNDROME IS DUE TO A PARTIAL DELETION OF THE SHORT ARM OF CHROMOSOME NUMBER 5, ALSO CALLED "5P MONOSOMY" OR "PARTIAL MONOSOMY." APPROXIMATELY 90% OF CASES RESULT FROM A SPORADIC, OR RANDOMLY OCCURRING, DE NOVO DELETION. THE REMAINING 10–15% ARE DUE TO UNEQUAL SEGREGATION OF A PARENTAL BALANCED TRANSLOCATION WHERE THE 5P MONOSOMY IS OFTEN ACCOMPANIED BY A TRISOMIC PORTION OF THE GENOME. THESE INDIVIDUALS MAY HAVE MORE SEVERE DISEASE THAN THOSE WITH ISOLATED MONOSOMY OF 5P. A RECENT STUDY SUGGESTS THIS MAY NOT BE THE CASE WHERE A TRISOMY OF CHROMOSOME 4Q IS INVOLVED

THERE IS NOT A SPECIFIC WAY TO TREAT THE CONDITION AS THE BRAIN DAMAGE CAUSED BY THIS CONDITION OCCURS IN THE EARLY STAGES OF EMBRYO DEVELOPMENT. INTENSIVE TREATMENT IS RARELY NEEDED IN INFANTS AND THEY CAN BE TREATED IN NEONATAL PATHOLOGY DEPARTMENTS. CHILDREN MAY BE TREATED BY SPEECH, PHYSICAL AND OCCUPATIONAL THERAPISTS. IF INFANTS HAVE DIFFICULTY IN SUCTION OR SWALLOWING, THEN PHYSICAL THERAPY SHOULD BEGIN IN THE FIRST WEEKS OF LIFE. HEART ABNORMALITIES OFTEN REQUIRE SURGICAL CORRECTION AND SPECIALIST ATTENTION [7]



DIAGNOSIS IS BASED ON THE DISTINCTIVE CRY AND ACCOMPANYING PHYSICAL PROBLEMS. THESE COMMON SYMPTOMS ARE QUITE EASILY OBSERVED IN INFANTS. AFFECTED CHILDREN ARE TYPICALLY DIAGNOSED BY A DOCTOR AT BIRTH. GENETIC COUNSELING AND GENETIC TESTING MAY BE OFFERED TO FAMILIES WITH INDIVIDUALS WHO HAVE CRI DU CHAT SYNDROME. PRENATALLY THE DELETION OF THE CRI DU CHAT RELATED REGION IN THE P ARM OF CHROMOSOME 5 CAN BE DETECTED FROM AMNIOTIC FLUID OR CHORIONIC VILLI SAMPLES WITH BACS-ON-BEADS TECHNOLOGY. G-BANDED KARYOTYPE OF A CARRIER IS ALSO USEFUL.



Diagnostic testing occurs when an individual is showing signs or symptoms associated with a specific condition. Genetic testing can be used to arrive at a definitive diagnosis in order to provide better prognosis as well as medical management and/or treatment options. Testing can reveal conditions can be mild or asymptomatic with early treatment, as oppose to debilitating without treatment (such as phenylketonuria). Genetic tests are available for a number of genetic conditions, including but not limited to: Down syndrome, sickle cell disease, Tay–Sachs disease, muscular dystrophy. Establishing a genetic diagnosis can provide information to other at-risk individuals in the family.

Any reproductive risks (e.g. a chance to have a child with the same diagnosis) can also be explored after a diagnosis. Many disorders cannot occur unless both the mother and father pass on their genes, such as cystic fibrosis; this is known as autosomal recessive inheritance. Other autosomal dominant diseases can be inherited from one parent, such as Huntington disease and DiGeorge syndrome. Yet other genetic disorders are caused by an error or mutation occurring during the cell division process (e.g. aneuploidy) and are not hereditary.