

THE DIVISION OF MEDICAL GENETICS

Royal University Hospital, Saskatoon, Saskatchewan

Pediatric Services

Genetic Counselling in pediatrics involves gathering information that might help establish a diagnosis. This includes evaluation of the prenatal, birth, developmental, medical and family histories, as well as physical examination of the child.

As a team, we anticipate the impact of a specific diagnosis for the family, and educate them about the results of our investigations and possible diagnosis. We make recommendations and referrals as necessary to ensure families have access to the most appropriate medical and social services. Genetic Counsellors are available for ongoing emotional support and to discuss family planning issues if necessary. Grief counselling can be initiated when a family is experiencing the loss of a child or following diagnosis.

Presymptomatic testing for adult onset conditions and carrier testing are not offered routinely to minors. Testing a minor fails to respect their future autonomy, breaches their confidentiality, and may cause psychological harm. Results of genetic testing could alter the family dynamic and bond between parents and children. Other harms include the potential feelings of inadequacy or loss of self-esteem for the young person tested; as well as the potential for discrimination when applying for life insurance, employment, or immigration.

Some examples of reasons to refer include children with birth defects, unusual appearance, developmental delay or regression of development, autism, speech delay, muscle problems, neurological abnormalities, unusual cancer diagnosis, endocrine abnormalities, hematological abnormalities, or a family history of a genetic or chromosomal disorder.

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