The Role of genetic counseling for chromosomal abnormalities:

Focus on parents of children who underwent submicroscopic chromosomal rearrengements investigation and parents identified as a balanced chromosomal translocation carrier

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Most of chromosomal abnormalities with gains or losses of genetic material are generally manifest the multiple congenital abnormalities, intellectual and/or developmental delay (MCA/ID•DD). Chromosomal abnormalities might be either numerical or structural. Numerical abnormalities of chromosome mean either losing one chromosome from a pair (monosomy) or having additional chromosomes (trisomy, tetrasomy, etc) and are generally manifest phenotypic effects. Structural chromosomal abnormalities, which may take various forms, can be classified into either unbalanced or balanced ones. Deletion or duplication of even a micro portion of chromosome could be meaningful unbalanced structural abnormality, which could lead to syndromic phenotype. On the other hand, balanced structural abnormality is local exchange of chromosome without gene dosage imbalance. Therefore, individuals who have balanced structural abnormality are called carriers and usually manifest no symptoms.

G-banded karyotyping is known as a chromosomal analysis method that can detect and diagnose all the numerical changes and rearrangements to some extent. For submicroscopic chromosomal rearrangements which are too small to detect by G-banded karyotyping, chromosomal microarray analysis (CMA) and Multiplex ligation-dependent probe amplification (MLPA) are used. Those
rearrangements, even being small are known to be the cause of up to 20% of MCA/ID·DD (multiple congenital anomalies and/or intellectual disability·developmental delay) because of submicroscopic chromosomal rearrangements for the present. CMA and MLPA show much higher diagnostic yield for accounting the unexplained ID/DD than G-banded karyotype that has been unable to do. CMA is proficient in detecting micro deletions and duplications of chromosomes comprehensively and MLPA is suitable for capturing deletions and duplications of specific gene region.

The small abnormalities detected by CMA or MLPA may be one of the following results: “detected no chromosomal imbalance”, “detected a mutation but disease relevance is unexplained”, or “detected a mutation which is likely to have disease relevance but uncertain clinically”. As the diagnostic significance being low, most of the parents of children with MCA/ID·DD, who underwent those genetic testings of CMA or MLPA are likely to receive the result of “no chromosomal imbalance was detected.” Lack of clear diagnosis, therefore, may evoke them a feeling of anxiety, guilty, and further mental fatigue to face the repeated uncertain diagnosis.

The first part of this study is focused on the parents of children who received negative test results of submicroscopic chromosomal rearrangements investigation. From the data obtained through interviews, we examined how the parents came to accept those results. To illustrate how parents came to understand the possible limitations, the sensitivity of the tests and their thoughts on undergoing the test and advancement of technology, they were asked with question as “How did they expect the tests to reveal something new?”, “How did they come to be satisfied with the results whatever the results they might receive?” and “What did they think of the results of no abnormality detected”. As the results, most of the parents, 89% (8/9 parents)
of them understood the tests correctly i.e. possible limitations, sensitivity of tests, and the methodologies used in analysis, through the genetic counseling session. Their 67% (6/9 parents) understood the significance of the test results. The parents at 78% (7/9 parents) were satisfied with CMA or MLPA results even though the result was “no chromosomal imbalance was detected”. They were also asked their thought on taking the test, and on advancement of technology. Most parents expected medical advances in future. They hoped for new testing to detect their situation more clearly and finally lead to “actually improve the quality of life of affected children”. At the same time they also had ambivalent feelings that they already felt like giving up in exploring a clear cut answer to their children’s diagnosis, and fear for discrimination against genetic variation.

The second study was focused on parents identified as a balanced chromosomal translocation carrier because they are at risk of having offspring with unbalanced structural abnormality. They might therefore experience the psychosocial problems such as anxiety, feelings of guilt, blaming themselves, or social stigmatization.

To study the psychosocial impacts of being a balanced chromosomal carrier that is found out through abnormal offspring, parents who had been identified as balanced chromosomal carriers were asked, such as psychosocial consequences after they heard the etiological explanation for the test, their plan of disclosing the information to their related family members, and their future reproductive plan. As the results, most of those who are identified as a balanced translocation carrier had correctly remembered what the term “carrier” means and the purpose of carrier testing. All the parents shared the information of their carrier status with their own parents and/or siblings. The parents realized and understood that the carrier status information of siblings
of affected children was very important for the siblings once they understood the meaning and influence to their reproductive decision. Thus, the parents wanted someone to discuss the issues on the disclosure of possible carrier status to unaffected children including how and when. The parents at 27% (3/11 parents) answered that they planed not to bear any other children and said that was not because of the carrier status of themselves.

This study had revealed various mixed and complex thoughts, emotion, and impacts in both parents of children who underwent submicroscopic chromosomal rearrangements with negative result and parents who were identified as balanced chromosomal translocation carriers. Our findings will be valuable for genetic counselors and other healthcare professionals before and after they offer the genetic testing. Health care professionals not only need to provide comprehensible description not using medical terms on the genetic testing they offer to enhance clients’ understanding, but also need to dig deeply into individuals’ expectation, needs, mental condition to support their better decision making. Because the result of genetic testing may relate to their family members, health care professionals also need to discuss feelings and emotions, which may occur, and to provide anticipatory guidance not only to clients but also to their family members before and after undergoing genetic testing.