

The Broad Autism Phenotype and the Quality of Life of Parents Who Have Children on the Autism Spectrum

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Abstract: This paper aims to study families who have children with autism spectrum disorder. The research interest was focused around the quality of life of these families, the factors affecting it and the occurrence of the autism phenotype in the relatives of children with the disorder. By studying the available literature, it became apparent that the autism phenotype in particular is a new area of research interest for the Greek data. For this reason, an attempt is made to study this phenomenon. In fact, it is of particular research interest to study the presence of the autism phenotype and the quality of the parents' friendships, how they may interact with the quality of life of the family. This study examines the broad autism phenotype and quality of life in parents of children on the autism spectrum and parents of children of typical development. 174 parents participated, from the Athens and Piraeus area with a mean age of 45.01 years (standard deviation = 7.9). Of the 174 parents, 119 were female (68.4%), and 55 were male (31.6%). The questionnaires were distributed to 99 parents who had children of typical development (56.9%) and 75 who had children on the autism spectrum (43.1%). The following questionnaires were used: a) the Greek version of the World Health Organization Quality of Life Questionnaire (WHOQOL), b) the Broad Autism Phenotype Questionnaire (BAP), c) the Friendship Questionnaire. The results showed that the presence of the autism phenotype is a significant predictor of poor quality of life and friendship for the parents. The autism phenotype was found to occur more frequently in parents who have children with autism. However, no significant differences in quality of life and friendship were found between parents with a child on the autism spectrum and parents of children of typical development.

Keywords: Broad Autism Phenotype, Quality of Life, Friendship

1. Foreword

This paper aims to study families who have children with autism spectrum disorder. The research interest was focused around the quality of life of these families, the factors affecting it and the occurrence of the autism phenotype in the relatives of children with the disorder. By studying the available literature, it became apparent that the autism phenotype in particular is a new area of research interest for Greek data. For this reason, an attempt is made to study this phenomenon. In fact, it is of particular research interest to study the presence of the autism phenotype and the quality of the parents' friendships, how they may interact with the quality of life of the family.

1.1. Autism Phenotype

The autism spectrum includes individuals who exhibit developmental disorders characterized by impaired social interaction, communication, stereotyped behavioral patterns and interests [27, 2]. Research has presented a cluster of genetic risk factors for the disorder [9, 3]. Indeed, it appears that the recurrence rate is somewhere between 5-8% within the family [23], and 60% in mono-zygotic twins and 3- 5% in zygotic twins [3]. In conclusion, autism is one of the most heritable complex psychiatric disorders. However, it appears that this genetic risk is shared by some of the relatives of individuals with autism, but without having the disorder. That is, they exhibit the autism phenotype [22, 24]. The study of the autism phenotype, or intra-phenotype and intermediate

phenotype, as reported by Carlson [5], shows that the focus shifts not to identifying a gene responsible for autism, but to presenting the disorder as a genetically complex psychiatric disorder. Intermediate phenotypes are referred to as heritable sub-clinical markers of the disorder, such as behavioral, physiological, neurobiological and other characteristics, which are present in both disorder-affected members and non-affected members of the same family [12].

The intermediate phenotype was first reported in a twin study by Folstein and Rutter [9]. Through the researchers' study, the cognitive and language skills of twins were studied, where only one member of the pair had the autism disorder. Language and cognitive deficits similar to his sibling were therefore identified in the typically developing twin. The existence of behavioral cognitive and language vulnerability markers of autism in members of the same family has therefore been recognized. It has also been found in many studies that in families with more than one member with the autism disorder, indicators of the intermediate phenotype will be more present than in families with one incident of autism [8].

1.2. *Quality of Life*

The birth and upbringing of a child with autism is a major event that completely changes a family's daily life. It is an unprecedented and puzzling situation that all family members have to face. The psychological, social, emotional and interpersonal strain is therefore intense. The majority of children with autism spectrum disorders live with their families. Caring for children can be demanding and can be accompanied by increased levels of stress [10]. Frank-Stromborg (1988), defines quality of life as the balance of an individual's psycho-social structure. This includes life satisfaction, happiness, cognitive balance, adjustment, functional status and health status. Caring for children with this disorder is associated with problems in cognitive and mental health also present higher levels of stress [26]. Parents with children on the autism spectrum experience feelings of intense anger, guilt, depression and anxiety. In fact, many times these emotional problems are expressed as psychosomatic symptoms, [26]. The daily life of the parent is quite difficult, as they are forced to follow their child in most activities. The parent's social life is further restricted and it is very difficult to change the family routine. Of course, parental stress and physical and mental health also depend on the characteristics of the child, the severity of the disorder, age and comorbidity with other disorders [11, 13].

1.3. *Theoretical and Empirical Evidence*

In reviewing the available foreign and Greek literature, no research was found that was identical to the present study, especially in terms of the co-examination of the factors of autism phenotype, quality of life and quality of parental friendship.

1.3.1. *Autism Phenotype/Quality of Friendship*

Regarding the autism phenotype, the literature that was found and studied was not from the Greek data. Researches

identify some specific characteristics in parents who display the autism phenotype, such as social reserve, distant personality and lower friendship quality. Furthermore, relatives who have the phenotype show less interest in their environment and difficulty in adapting [21, 18]. Moreover, regarding the language skills of these individuals, there is no easy flow in narrative and sequencing of thought [22]. Through the study of previous research it was shown that the autism phenotype occurs in parents who have children on the autism spectrum. Researchers emphasize the gene effect of the disorder, which is expressed in the children's relatives as a phenotype [24, 20]. In addition, since these parents have characteristics that make social contact difficult, such as distant personalities, rigid behavior and reduced language skills, they show a lower quality of friendship [21, 18].

1.3.2. *Quality of Life*

Regarding the quality of life of parents, a variety of research has been studied in both Greek and foreign literature. In summary, the majority of studies agree that parents of children on the autism spectrum suffer from high stress [15], depression [6], and a general negative mood [1]. In addition to mental stress, parents also experience reduced physical functioning, fatigue and exhaustion, [14, 7]. In fact, Weiss [26], studying the effects of intense anger, guilt and depression in parents, concluded that because of these emotional problems, psychosomatic symptoms often develop.

1.4. *Purpose of This Research/Research Hypotheses*

By examining the available Greek literature, it quickly became clear that there are no published studies dealing with the occurrence of the autism phenotype in Greek families. The aim of the present research is to study the phenomenon of the broader autism phenotype and its correlation with the quality of life and friendship of the parents. Thus, an attempt is made to extend the existing knowledge about the occurrence of the autism phenotype in Greek families. After studying the literature data and relevant articles and research, the following research hypotheses were formulated regarding the study of families with children on the autism spectrum.

1st research hypothesis:

Parents who have children on the autism spectrum will show lower quality of life and quality of friendship.

2nd research hypothesis:

Autism phenotype will be more frequently identified in parents who have children on the autism spectrum.

3rd research hypothesis:

In parents whose the autism phenotype can be identified, regardless of whether they have a typically developing child or a child on the autism spectrum, will have a lower quality of life and friendship.

2. *Method*

2.1. *Participants*

174 parents (n=174), from the Athens and Piraeus area,

with a mean age of 45.01 years (standard deviation = 7.9), participated in the study. Of the 174 parents, 119 were female (68.4%), and 55 were male (31.6). Regarding their educational level, the majority were high school graduates (45.4%), followed by graduates of universities. (27.6%), technical education institutions (8.6%), Master's degree holders (8%), high school graduates (6.9%) and PhD holders (2.3%). Regarding marital status, the majority of participants were married (82.8%). Regarding the experimental and control groups, 99 parents who had typically developing children (56.9%) and 75 who had children on the autism spectrum (43.1%) participated. The parents with typically developing children came from a primary school in Korydallos, while the parents with children on the autism spectrum participated from various medical and educational centers and special schools in Athens and Piraeus.

2.2. Research Tools

Three questionnaires were used to collect data, in addition to the demographic characteristics questionnaire:

- 1) Quality of Life Questionnaire, World Health Organization. This questionnaire is the translated version of the quality of life world health organization (WHOQOL). The Greek translation and editing was done by Kokkosi, Antonopoulou and Christodoulou. It is a questionnaire of 26 statements assessed with five Likert-type scales (1=never, 2=rarely, 3=sometimes, 4=very often, 5=always). The questionnaire contained 4 sub scales. The sub scale of Physical Health, Psychological Health, Social Relationships and Environment. At the beginning of the questionnaire there are two questions about the perceived physical and mental health of the participants not belonging to the sub scales. The Cronbach's internal consistency coefficient alpha (a) was 0.933, which is quite high and confirms the reliability of the adaptation of this questionnaire to the Greek population.
- 2) Questionnaire of autism phenotype. This is the translated version, which was made by the researcher, of the Broad autism phenotype questionnaire (BAP) (Hurley R. S., Losh, Parlier, Reznick, & Piven, 2007). It is a 36 statement questionnaire rated on a 6-point Likert-type scale (1=very rarely, 2=rarely, 3=sometimes, 4=somewhat often, 5=often, 6=very often). This instrument consists of three sub scales that identify three

main characteristics of individuals with the autism phenotype. These are aloof personality defined as a lack of interest or enjoyment of social interaction, rigid personality which characterizes individuals with little interest in change or difficulty in adapting, and pragmatic language which relates to deficits in communicative language skills, in resolving communication difficulties effectively, in maintaining a flow in conversation and in mutual discussion. The Cronbach's internal consistency coefficient alpha (a) reliability was 0.917, which is quite high and confirms the reliability of the adaptation of this questionnaire to the Greek population.

- 3) Friendship Questionnaire. It is the translated version, carried out by the researcher, of the Friendship Interview used by Losh and Piven in their study on the autism phenotype (Social-cognition and the broad autism phenotype: identifying genetically meaningful phenotypes, 2007). In this measure, participants were asked to mark three friends outside the family environment. The quality of these friendships is assessed through 6 questions that can be scored as 0-15. Previous research has shown how this instrument is useful in assessing social relationships and in distinguishing parents with children on the autism spectrum from parents in the control group [22]. Cronbach's internal consistency coefficient alpha (a) reliability was 0.855, which is quite high and confirms the reliability of the adaptation of this questionnaire to the Greek population.

3. Results

3.1. Descriptive Analysis

Table 1 shows the difference between the means of parents who do not have children with autism ($M=3.71$ t.a.=0.53), with parents who have children with autism ($M=3.38$ t.a.=0.62), in terms of quality of life where it is not statistically significant ($t_{172}=3.705$, $p<0.051$). Furthermore, the differences in means of the two groups mentioned above are also presented with regard to the sub scales of the questionnaire (physical health, psychological health, social relations, environment). Statistical differences are found in the sub scale of psychological health and social relations, where it appears that parents who do not have children with autism do better in these areas.

Table 1. Comparison of means of quality of life and its sub scales (physical health, psychological health, social relationships, environment) of parents with typically developing children and parents with children on the autism spectrum.

	Parents with typical developing children (n=99)			Parents with children on the autism spectrum (n=75)		
	Mean	Standard Deviation	Mean	Standard Deviation	t-test	p
Quality of life	3,71	0,533	3,38	0,620	$t_{172}=3,705$	0,051
Physical Health	3,91	0,686	3,54	0,700	$t_{172}=3,518$	0,427
Psychological Health	3,65	0,524	3,38	0,616	$t_{172}=3,111$	0,036
Social Relationships	3,97	0,757	3,55	0,948	$t_{172}=3,082$	0,015
Environment	3,40	0,656	3,16	0,651	$t_{172}=2,457$	0,783

Table 2 shows the difference in means of parents with typically developing children ($M=2.63$ t.a.=0.451) and parents

with children on the autism spectrum ($M=2.99$ t.a.=0.769) in terms of autism phenotype, which was found to be statistically

significant ($t_{172}=-3.594$, $p=0.001$). That is, parents who have children on the autism spectrum will have a higher frequency of the autism phenotype than parents who have children with typically developing children. Furthermore, Table 2 will also present the differences in the means of these groups with respect to the sub scales of the autism questionnaire (aloof

personality, realistic language, rigid personality). Statistically significant differences appear with respect to distant personality and realistic language and not in the sub scale of rigid behavior. Thus, it appears that parents with children on the autism spectrum exhibit the above items more frequently than parents with typically developing children.

Table 2. Comparison of the means of the autism phenotype and its sub scales (distant behavior, realistic language, rigid personality) of parents with typically developing children and parents with children on the autism spectrum.

	Parents with typical developing children (n=99)			Parents with children on the autism spectrum (n=75)		
	Mean	Standard deviation	Mean	Standard deviation	t-test	p
Autism Phenotype	2,63	0,451	2,99	0,769	$t_{172}=-3,594$	0,00
Aloof personality	2,54	0,613	3,05	1,001	$t_{172}=-3,855$	0,00
Realistic language	2,13	0,503	2,61	0,790	$t_{172}=-4,630$	0,00
Rigid personality	3,22	0,658	3,30	0,744	$t_{172}=-0,734$	0,095

Table 3 presents the differences between the means of parents with typically developing children ($M=11.49$ t.a.=2.76) and parents with children on the autism spectrum ($M=8.64$ t.a.=3.30) in terms of friendship quality, where the difference is not

statistically significant ($t_{170}=6.145$, $p=0.085$). In other words, it appears that both parents who have children on the autism spectrum and parents who have typically developing children do not differ in the quality of friendships they create.

Table 3. Comparison of means of friendship quality of parents who have typically developing children and parents with children on the autism spectrum.

	Parents with typical developing children (n=99)			Parents with children on the autism spectrum (n=73)		
	Mean	Standard deviation	Mean	Standard deviation	t-test	p
Friendship quality	11,49	2,775	8,64	3,297	$t_{170}=6,145$	0,085

3.2. Associations

The correlation analysis, as shown in Table 4, revealed statistically significant correlations between the autism phenotype of parents with typically developing children and their quality of life and friendship. Specifically, there was a negative correlation between autism phenotype and quality of life ($r=-0.514$, $p<0.001$) and quality of friendship ($r=-0.413$, $p<0.001$).

Table 4. Association between autism phenotype of parents with typically developing children (n=99), and quality of life and friendship.

	Autism phenotype	p
Quality of life	-0,514**	0,00
Friendship quality	-0,413**	0,00

* $p<0,05$, ** $p<0,01$, *** $p<0,001$.

Table 5 presents the correlation analysis where statistically significant differences were found between the autism phenotype of parents with children on the autism spectrum and their quality of life and friendship. There was a negative correlation between autism phenotype and quality of life ($r=-0.772$, $p<0.001$) and quality of friendship ($r=-0.497$, $p<0.001$).

Table 5. Association between autism phenotype of parents with children on the autism spectrum (n=75), and their quality of life and friendship quality.

	Autism phenotype	p
Quality of life	-0,772**	0,00
Friendship quality	-0,497**	0,00

* $p<0,05$, ** $p<0,01$, *** $p<0,001$

3.3. Regression Analysis

In Table 6, autism phenotype explained 77.2% of the variance ($R^2=0.596$, Adjusted $R^2=0.590$) in terms of quality of life of parents with children on the autism spectrum ($F_{1,73}=107.60$, $p<0.001$). Analytically, autism phenotype predicted quality of life at a statistically significant level ($\beta=-0.622$, $t=-10.37$, $p<0.001$). Furthermore, a regression analysis was performed where autism phenotype was set as a predictor for parents who have typically developing children. Specifically, autism phenotype explained 51.4% of the variance ($R^2=0.264$, Adjusted $R^2=0.257$), in terms of quality of life for parents with typically developing children ($F_{1,97}=34.88$, $p<0.001$). In detail, autism phenotype predicted at a statistically significant level the quality of life of parents with typically developing children ($\beta=-0.609$, $t=-5.906$, $p<0.001$).

Table 6. Regression analysis for quality of life of parents with children on the autism spectrum ($N=99$) and parents with typically developing children ($N=75$) by autism phenotype.

Variance	B	SE	β	Variance	B	SE	β
Parents with children on the autism spectrum	-0,62	0,60	-0,77***	Parents with typical developing children	-0,61	0,10	-0,51***
R2	0,60***			R2	0,26***		
Adjusted R2	0,59***			Adjusted R2	0,26***		

* $p<0,05$, ** $p<0,01$, *** $p<0,001$.

In Table 7, autism phenotype explained 49.7% of the variance ($R^2=0.247$, Adjusted $R^2=0.236$) in terms of the quality of friendship of parents with children on the autism spectrum ($F_{1,71}=23.302$, $p<0.001$). Analytically, autism phenotype predicted friendship quality at a statistically significant level ($\beta=-2.111$, $t=-4.827$, $p<0.001$). Furthermore, a regression analysis was performed where autism phenotype was set as a predictor for parents who

have typically developing children. Specifically, autism phenotype explained 41.3% of the variance ($R^2=0.17$, Adjusted $R^2=0.162$), in terms of friendship quality of parents with typically developing children ($F_{1,97}=19.891$, $p<0.001$). Analytically, autism phenotype predicted at a statistically significant level the quality of friendship of parents with typically developing children ($\beta=-2.541$, $t=-4.460$, $p<0.001$).

Table 7. Regression analysis for friendship quality of parents with children on the autism spectrum ($N=99$) and parents with typically developing children ($N=73$) by autism phenotype.

Variance	B	SE	β	Variance	B	SE	β
Parents with children on the autism spectrum	-2,11	0,44	-0,50***	Parents with typical developing children	-2,54	0,57	-0,41***
R2	0,25***			R2	0,17***		
Adjusted R2	0,24***			Adjusted R2	0,16***		

$p^*<0,05$, $p^{**}<0,01$, $p^{***}<0,001$.

4. Discussion

The main purpose of this research is to investigate the occurrence of the autism phenotype in Greek families and how it affects the quality of life of parents. In combination, the quality of friendships that parents establish and how this is affected by the presence or absence of the autism phenotype is examined. The ultimate goal of this research is to expand the information regarding this phenomenon, since it appears quite limited in the Greek literature. Thus, we hope to be able to provide more effective help and support to families who have children on the autism spectrum by now considering the sensitivity of parents who present the phenotype.

The results of the study do not confirm the initial hypotheses that parents who have typically developing children will have a better quality of life than parents who have children on the autism spectrum. However, in terms of the sub scales of quality of life, it appeared that parents with typically developing children have better psychological health and have a more positive perception of their social relationships. These findings are also consistent with findings from previous studies that suggest that parents with children on the autism spectrum have increased rates of depression, higher levels of anger and general psychological difficulties in their daily lives ([28, 4]. These difficulties are directly linked to children's behavioral problems and hyperactivity, as well as their reduced participation in social activities, which affects the parents' own sociability, which is shown to be reduced. However, the results differ from the research [14] regarding physical functioning and perception of the environment in parents with children on the autism spectrum, as no significant differences were found between parents with typically developing children.

Regarding the autism phenotype, it was found how it occurs more frequently in parents with children on the autism spectrum, confirming the original research hypothesis. Specifically, parents who have children on the autism

spectrum were found to be more distant and use more pragmatic language where they find it difficult to communicate comfortably and maintain a flow in the conversation compared to parents who have typically developing children. These findings are consistent with previous research [24]. No difference was found in terms of parental personality rigidity and adjustment difficulty between parents with children with autism and parents with typically developing children. Furthermore, no significant differences were found in the quality of the friendships formed between the two groups, thus rejecting the original research hypothesis and showing different results from previous research [11, 13]. However, as will be discussed below, it appears that the presence of the phenotype is a stronger predictor of lower friendship quality than having a child with autism in the family.

The association between autism phenotype and quality of life appeared to be negative for both parents with children on the autism spectrum and parents with typically developing children. Thus, parents with the autism phenotype have a lower quality of life and also a lower quality of friendships. As already mentioned, confirming the research hypothesis, the strongest predictor of parents' quality of life and friendship quality is the presence of the autism phenotype, regardless of whether parents actually have children with autism or typically developing children. It seems, therefore, that the existence of unexpressed autism traits in parents makes them more vulnerable with regard to the quality of the friendships they establish and their quality of life. Thus, characteristics such as social reserve, aloofness, difficulty in adapting and little interest in their environment affect the quality of their daily life and the friendships they form [21]. It can be assumed that having children on the autism spectrum further impairs the quality of life and the quality of friendships of these parents.

5. Conclusion

In summary, the present study highlights the relationship

between the autism phenotype and the quality of life and quality of friendships of parents. It appears, therefore, that when attempting to support families with children on the autism spectrum, many factors need to be controlled for before deciding on the most effective and appropriate course of intervention. As it was shown by this research, the appearance of the autism phenotype in parents affects the outcome of any efforts of the specialists who will deal with the child. In particular, it seems that parents must be included in this multifaceted intervention, not only for reasons of emotional support, but also because they themselves must be the target of intervention techniques in order to manage the specific non-functional characteristics that appear in the autism phenotype.

6. Limitations

A key limitation of the study, was perhaps the unsatisfactory sample of families with children on the autism spectrum, which influenced the results, and this is probably why there was no agreement, in terms of the quality of life and friendship of parents with children on the autism spectrum, with the research hypotheses and with previous research. Of course, because the investigation of the phenomenon of autism in Greek families is limited and there is no abundance of research material, further scientific work and research on this topic will bring more certain results.

7. Suggestions for Further Research

Some thoughts on further advancing this research topic are that questions about the sources of support received by parents who have children on the autism spectrum could be added to the questionnaire. Specifically, whether they have been to medico-educational centers, whether they are supported by different groups, what is the relationship with their partner, friends and more. By studying the literature, it seems that support from different structures and quality relationships are protective factors and have a positive impact on the quality of life of parents with children on the autism spectrum [19, 16]. It would be of particular interest to also examine the personality of parents and identify specific characteristics of this personality that may be protective or risk factors for their quality of life [25]. Furthermore, the study of families with more than one child with autism appears quite frequently in foreign literature. It seems that the autism phenotype occurs more often in these families than when there is only one child with autism [17]. A relevant study could therefore be carried out for the Greek data.

8. Restrictions

A key limitation of the study was perhaps the unsatisfactory sample of families with children on the autism spectrum, which influenced the results and is likely to be why there was no agreement, in terms of quality of life and friendship of parents with children on the autism spectrum, with the research hypotheses and with previous research. Of

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